NTSAD Community News

Research, Collaboration, and Community

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

January

January 2024

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ONE MONTH AWAY: Raise Your Voice to Advance GM2 Research

The biggest opportunity yet for the GM2 Community to share their rare experiences is ONE MONTH AWAY! **The first and only Externally-Led Patient-Focused Drug Development (EL-PFDD) Meeting for GM2 (Tay-Sachs or Sandhoff diseases) is on February 15, 2024, 9:30 a.m. Eastern.** Hosted by NTSAD, the meeting is held virtually and livestreamed via YouTube. All are welcome – families, researchers, clinicians, members of industry, et. al.

Registration is required.

Even if you are not personally affected by Tay-Sachs or Sandhoff disease, consider joining the meeting in support of rare families and your fellow NTSAD Community members.

The EL-PFDD meeting will give the GM2 Community a chance to share their experiences and perspectives with members of the U.S Food and Drug Administration (FDA) and other key stakeholders, including medical product developers, health care providers, and federal partners. It is an opportunity for them to learn about the symptoms that matter most, the impact the disease has on daily life, and community member's personal experiences with currently available treatments. This input can inform the FDA's decisions and oversight both during drug development and during review of a marketing application. *It is an incredible opportunity to advocate and speak directly to those who can drive progress forward for GM2!* And it's free and easy to attend!

If you want to learn more about the EL-PFDD meeting, watch the Community Informational Webinar to hear what to expect and how you can participate. Don't forget to register!

Register for GM2 EL-PFDD now!

Share Your Experience in Advance of the GM2 EL-PFDD Meeting

Are you or someone you love personally affected by GM2 (Tay-Sachs and Sandhoff disease)? Prior to the GM2 EL-PFDD meeting, you or a caregiver can submit comments and

information about your experience with the disease. **Your comments may be shared during the GM2 EL-PFDD meeting and may also be used in the final Voice of the Patient summary report.** (All identifying information will be removed). Lend your insight and help us advance research and find treatments!

Submit an early comment.

Share your GM2 experience

NTSAD's Annual Family Conference 2024

The NTSAD Community is headed to the windy city for the 46th Annual Family Conference! We hope you will join us April 11 to 14, 2024 in Chicago, Illinois, for four days of connection, collaboration, and support. The conference is a safe place that brings rare individuals and families together and where they can meet with researchers, patient advocates, and industry members, too. All are welcome! Last June more than 300 people attended the conference. Save your place and **register today!**

Register for the Annual Family Conference

Don't forget to reserve your hotel room at the Conference.

If you're an individual or family coping with a diagnosis of Tay-Sachs, Canavan, GM1, or Sandhoff disease and in need of financial assistance to attend the conference, you can apply for a Helping Hand Grant.

Apply for a Helping Hand Grant

Sponsorship makes it possible for NTSAD to hold an unforgettable experience for our rare community. **Learn more about the sponsorship opportunities here.**

Sponsor the Conference

Thank you to 2024 Conference Sponsors!

Hope

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Susan* and Alan Roden and Family

Sponsor listing as of print date.
**NTSAD Board Member*

February is Rare Disease Awareness Month

Leading up to Rare Disease Awareness Month, we are asking-- what is Rare to you?

Is Rare Strong? Brave? Empowering? Hard? Beautiful? Inspiring?

How do you advocate for Rare? Why is raising rare voices important to you?

Email NTSAD Development and Communications Manager, Sydnie Dimond at sdimond@ntsad.org and share your thoughts on what "Rare is," what rare means to you, and a photo of you or your rare loved one. We will share your experience as together we advocate for rare on NTSAD's social media and in our newsletter during the month of February. We want to hear YOUR rare voice!

Share - What Rare Means to You

NTSAD and Uplighting Athletes Give Grant for GM2 Research

Toloo Taghian, PhD alongside nine other early-career researchers have been selected as a part of the 2024 Uplifting Athletes Young Investigator Draft on February 3 for her GM2 research.

Uplifting Athletes' Young Investigator Draft is inspired by the NFL Draft but shifts the focus from the selection of potential talent on the football field to recognizing the next generation of promising young medical researchers in the rare disease space.

NTSAD nominated Dr. Taghian and is matching her Uplifting Athletes' $10,000 grant, for a total of $20,000, to support development of gene therapy for Tay-Sachs and Sandhoff diseases at the University of Massachusetts Chan Medical School.

Learn more.

Katie & Allie Buryk Research Fund Surpasses $1 Million Goal

In 2014, after searching for a diagnosis for eight years, Katie Buryk finally had one-- Late Onset Tay-Sachs disease. She and her twin sister Allie are both affected by the ultra-rare genetic disorder, that leads to muscle weakness in the legs, difficulty with speech and swallowing, and other progressive and life-altering symptoms.

When Katie received the diagnosis, she understood that there were no treatments or cures. But she and her family decided to do all they could to advance research. When launching the Katie & Allie Buryk Research Fund a decade ago, Katie shared what motivates her,


We Have Late Onset Tay-Sachs...
Do You Know What It Is?

Allie & Katie Buryk's Story

It took the two of us, 28-year-old twins, eight years to find out what was wrong.

Our symptoms started slowly. At first, there was the difficulty climbing stairs. Our leg muscles were weak and we had trouble standing from a sitting position without using our hands. We couldn't squat. Sometimes our legs would just give out and we would fall. And our speech changed, words came tumbling out and sometimes people didn't understand us. Finally, genome sequencing informed us that we had Late Onset Tay Sachs. And now here it is two years later, with no treatment and no cure.

Allie (left) & Katie Buryk.

"I have decided to go public and try to raise money, awareness and mount a search for a cure. Sure, it's for me and Allie, but it is also for infants and kids who aren't as lucky as we are. And it's for the families who must embark on this difficult, sad and baffling journey."

To date, the Buryk Research Fund has raised $1,076,667 for research for Late Onset Tay-Sachs as well as Sandhoff diseases with the help of more than 2,400 donors. You may have seen the Buryk Family's ad which runs quarterly in *The New York Times*. Thank you to all the donors who were moved by Katie and Allie's story and made a donation. It's because of you that the Buryk Research Fund is able to fund research, host six Annual Think Tanks to foster collaboration and innovation, and one day, soon find effective treatments for Late Onset Tay-Sachs and Sandhoff diseases.

Support the Buryk Research Fund

NTSAD Welcomes New Research Associate

This month Cyndy Perreault-Micale, PhD, joined NTSAD as our Research Associate. Cyndy is an accomplished scientist with extensive experience in basic molecular biology research, science writing and editing, genetic testing, and data analysis. Through her work at genetic testing companies Sema4/GeneDX and GoodStart Genetics, she was inspired to join the field of rare diseases and advocate for people affected by them. At NTSAD, Cyndy will manage scientific working groups; support an international network of researchers, clinicians, and industry partners; and help drive NTSAD's Research Initiative.

The Impactful Relationships between Rare Families and Researchers

As the NTSAD Community knows, rare families have been advancing research for decades. Affected families bring resources, funding, connections, and most importantly, their personal stories and experiences into the labs. The scientists at UMass Chan Medical School deeply value their partnerships with families, for the path forward is brighter when working together, and when the research affects a child and family they have come to know well.

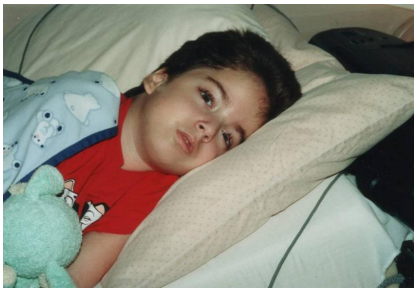
Read "They are the heroes" article.

Mathew Forbes Romer Foundation Honors Kathy Flynn

Each year at the Mathew Forbes Romer Foundation (MFRF)'s "Sweetness and Laughter" Celebration, an individual is honored for their impact on the rare community. This year, MFRF has chosen to honor NTSAD's Chief Executive Officer, Kathy Flynn, for leading the implementation of the Externally-Led Patient Focused Drug Development (EL-PFDD) Meeting for GM2 (Tay-Sachs & Sandhoff). The Mathew Forbes Romer Foundation is a sponsor for the EL-PFDD meeting, which is on February 15, 2024, and members of the FDA and key stakeholders also will be attending.

Learn more about the MFRF's "Sweetness and Laughter" Celebration.

The Mathew Forbes Romer Foundation
SINCE 1998
EDUCATION & RESEARCH OF CHILDREN'S GENETIC DISEASES OF THE BRAIN



What Matters Most Study: Help to Improve Pediatric Palliative Care

WHAT MATTERS MOST

Identifying a Core Indicator Set for Quality Pediatric Palliative Care

How do we know that the palliative care provided to children living with serious illness and their families is the best it can be? How do we keep improving?

First, we need to identify the most important criteria or "indicators" of high-quality pediatric palliative care.

There are more than 100 different indicators that could be used. We need children living with serious illness, their families, and healthcare professionals to take part in our study and help us pick the most important ones.

Join one of our panels to help!

The Family Panel includes children with a serious illness, families of children with a serious illness, and families of children who passed away from a serious illness.

The Healthcare Professional Panel includes clinicians, researchers, and administrators who are involved in palliative care for children.

Panel members in this study will complete three surveys over the next eight months to agree upon and rank the most important indicators of quality pediatric palliative care.

Visit our Website to Join a Panel: kimwidger.ca/what-matters-most/

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Learn more about the What Matters Most Study.

If you are interested in participating in this study, please email whatmattersmost@utoronto.ca to request your link to begin the first survey. Study close date: January 26, 2024

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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