NTSAD Community News

Research, Collaboration, and Community

Supporting families is the center of everything we do…

August

August 2023

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NTSAD Will Host First-Ever, Externally-Led GM2 PFDD Meeting

On February 15, 2024, members of the GM2 (Tay-Sachs and Sandhoff diseases) community will gather for the first-ever, Externally-Led Patient-Focused Drug Development (EL-PFDD) meeting to share their perspectives living with these rare diseases with the members of the U.S. Food and Drug Administration (FDA) to ensure that their experiences, needs, and priorities are captured and meaningfully incorporated into drug development and evaluation. **The FDA considers members of the GM2 community "experts in what it is like to live with their condition, and they are uniquely positioned to inform the understanding of the therapeutic context for drug development and evaluation."**

Members of industry and people interested in GM2 are encouraged to register to attend and watch the meeting in real time or the recorded version. The EL-PFDD meeting will be open to the general public and will air live on NTSAD's website. Following the meeting, it will be available to watch at anytime on NTSAD's YouTube channel.

Save the Date: February 15, 2024, for the GM2 EL-PFDD meeting.

Canavan FDA Listening Session Summary Now Available

On March 21, 2023, several parents whose children are or were affected with Canavan disease shared their experiences during a Patient-Led Listening Session with the FDA, so its

Gottlieb Scholarship for Rare Siblings

members could learn more about the symptoms, onset levels, and challenges in getting a diagnosis, as well as the complex care needed for a child with a rare disease and its impact on the child and entire family.

Thank you to the parents who shared stories about their loved ones and their personalities, expressed their hopes for their children, and advocated for the urgent need for effective treatments. This listening session was a collaborative advocacy effort by NTSAD, Canavan Foundation, and Cure Canavan Fund.

Read Canavan Diseases FDA Listening Session Summary.

Read more about NTSAD's FDA Engagement.

Show that You Care for Rare, Always

August is National Make-A-Will Month. Many families have included NTSAD in their wills to honor loved ones and to add to their legacies.

In 2018, Michael Sussman and Renée Licht established the Evelyn and Leonard Sussman Legacy Circle at NTSAD with a generous gift to recognize the remarkable and lasting contributions of Michael's parents, Evelyn and Leonard, who along with other families founded NTSAD in 1957. Today, the Sussman Legacy Circle honors those families who have included NTSAD in their estate plans.

If you are considering drafting or updating a will, **you can honor a loved one and ensure that NTSAD is always here for families needing support by contacting your estate attorney or financial advisor and asking to include a gift to NTSAD.** It is as simple as adding a codicil, or addendum, to your current will that states you wish to designate a gift of a certain amount or a percentage of your estate to NTSAD.

If you have already included NTSAD in your estate planning, contact NTSAD's Director of Development and Communications, Susan Keliher at 617-277-4463 or at **skeliher@ntsad.org,** so that we may thank you and include your name in future Sussman Family Legacy Circle publications.

In Michael and Renée's own words, ***"What began in the living rooms of a handful of New York families grew into the powerful and reputable national organization that exists today. We've established the Evelyn and Leonard Sussman Legacy Circle to recognize NTSAD's heritage and to honor Evy and Lenny's service, to perpetuate their memory, and to strengthen efforts toward finding new treatments and a cure for these devastating diseases."***

Imagine & Believe 2023

SAVE the DATE: November 9, 2023
6-9 pm, Royal Sonesta Boston

Jayne Gershkowitz
Chief Patient Advocate, Amicus Therapeutics
2023 NTSAD Honoree

CONNECTION. COLLABORATION. COMMUNITY.

Imagine & Believe brings together families, industry members, clinicians, researchers, and rare allies to envision a world with effective treatments for Tay-Sachs, Canavan, GM1, and Sandhoff diseases. **Join us on Thursday, November 9, at the Royal Sonesta Boston for NTSAD's signature event.** Your support of ***Imagine & Believe*** helps families and raises money for programs and research.

At the event, a rare family will share their inspiring story. **We also will honor Jayne Gershkowitz, Chief Patient Advocate at Amicus Therapeutics and former NTSAD Executive Director, for her trailblazing leadership in patient advocacy and championing children, adults, and families living with rare diseases.** Join us as we pay tribute to Jayne and thank her for a lasting impact on NTSAD and the rare community.

Admission begins at $200 per guest, and sponsorship opportunities range from $500-$25,000.

View sponsorship opportunities.

Reserve your place at Imagine & Believe.

Read more about the event.

Questions contact Sydnie Dimond, Development and Communications Manager, at 617-277-4463 or at **sdimond@ntsad.org.**

Sponsor, Purchase Admission, or Donate Here

Thank you to our 2023 *Imagine & Believe* sponsors, who make it possible for NTSAD to provide support services and programming for families and fund critical research.

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Amicus Therapeutics, Inc.

HOPE SPONSORS
Michael and Caitlin Gladstone
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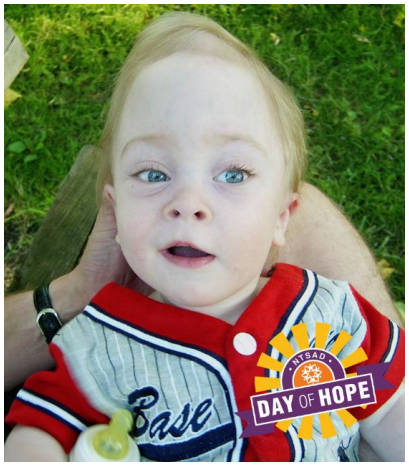
DETERMINATION SPONSOR
Staci Kallish*

*NTSAD Board Member

If you are interested in sponsoring, please contact NTSAD's Director of Development and Communications, Susan Keliher at **skeliher@ntsad.org.**

Sponsors as of publication date.

The Gropp Family's Day of Hope: *Sweet Brodryck's Legacy*



Each year, the Gropp Family hosts a Day of Hope event in honor of Brodryck, who had GM1 gangliosidosis. On August 5, they gathered their family and friends for a beanbag (cornhole) tournament, a raffle, and potluck dinner. NTSAD is grateful for the Gropp Family's efforts to raise awareness and funds for research.

"We thank everyone so much for their hard work, kindness, and dedication to our Sweet Brodryck's Legacy. It means the world to us that so many people have taken the time to remember and honour our Brodryck. This money goes directly into research with hopes that we can find that breakthrough. ***Hope is on the horizon that a cure will be found, and no other child will die from this dreaded disease."*** -Craig, Sherri, and daughter, Honor Gropp

There are many ways to participate in this year's Day of Hope, September 9 or a day of your choice. You can host your own event like the Gropps did or join the NTSAD community and Move a Mile for Hope. Learn more about Day of Hope.

Give to Day of Hope and Support Research Today!

Amber Franzen Joins NTSAD Board of Directors

NTSAD recently welcomed Amber Franzen as our newest Board member. When Amber's son, Rex, was diagnosed with infantile Sandhoff disease, Amber connected with NTSAD and found a rare community who truly understood what her family was going through.

"I am excited to join the NTSAD Board and see this as a way to become more involved in an organization that means so much to me and my family. I look forward to contributing and giving back to the NTSAD community." -Amber Franzen

Amber will be offering her knowledge as a rare mom to our Family Services Committee and her financial expertise to our Finance Committee. For many years, Amber has been a part of the Annual Family Conference Commemoration Committee, which plans the annual tribute for loved ones of conference attendees who have passed from Tay-Sachs, Canavan, GM1, and Sandhoff diseases.

Be a Rare Advocate: Making Expanded Carrier Screening More Equitable

September marks three National Awareness Month Campaigns — for Tay-Sachs disease, Leukodystrophy, and Newborn Screening — providing multiple opportunities for you to advocate for rare. A vital part of spreading awareness is prevention. Expanded Carrier Screening (ECS) gives families the information they need to safely plan and expand their families. In the video below, NTSAD Family Services Manager and Mom to Miss Elliott, Becky Benson, shared her story and presented on the importance of creating equity by providing coverage for ECS to **Washington State Healthcare Authority** on behalf of NTSAD and the **Access to Equitable Carrier Screening Coalition (AECS)**.

You can join Becky and NTSAD in this initiative by writing to your state Medicaid program to request they cover ECS, so that everyone has access to this important diagnostic

tool. **Use NTSAD's letter template. Learn more about AECS and how to write your letter.**

NTSAD understands how vital ECS is for family planning, especially for families who already know they could be potential carriers due to having a relative affected by a rare disease. As such, NTSAD offers free comprehensive carrier screening services to family members of an affected child or individual. hanks to Canavan Foundation, the Evan Lee Ungerleider Fund of NTSAD, Mathew Forbes Romer Foundation, and the NTSAD New York Area Fund, for making free screening possible through JScreen. Go to **JScreen.org** and use code NTSAD23 for a ReproGen test at no cost to you.

Looking for more ways to get involved and raise awareness this September?

Follow NTSAD on **Facebook,** **Instagram,** **LinkedIn,** and **Twitter** and share our awareness posts!

Participate and support NTSAD's Day of Hope, which raises awareness and money for research initiatives. You can also Move a Mile for Hope on September 9. Learn more.

Global Gene's RARE Advocacy Summit Experience

One of the world's largest gatherings of the rare community, Global Gene's RARE Advocacy Summit, Global Gene's RARE Advocacy Summit, is September 19-21, at the Sheraton San Diego Hotel & Marina on Harbor Island Drive in San Diego. CEO Kathy Flynn will attend this conference to represent NTSAD.

Learn more about the Summit.

Last fall, NTSAD's Family Services Manager and Mom to Miss Elliott, Becky Benson, attended Global Gene's RARE Advocacy Summit last fall and participated in a panel discussing grieving as a community. Becky recently shared this experience in a blog post she wrote for Global Genes.

"I came away from each session I attended with pages of notes to call upon in my support work, and new colleagues with whom I could connect. Most importantly, it provided a space where I could share my daughter with others while continuing to expand on her legacy and honor her all-too-short life. ***The ability to see individuals for who they are, apart from their diagnosis, and understanding the impact we can all have in this world is imperative to our humanity as a whole.*** *At the RARE Advocacy Summit, we are all one, working together to serve this purpose."* -Becky Benson

Read the full blog post.

Summer Intern Meghan Werner

This summer, Meghan Werner, served as a research intern at NTSAD. Meghan, a rising senior at Clemson University, who is pursuing a Bachelor of Science degree in genetics, with a minor in Spanish, supported several strategic projects.

Caroline Aragón, MS, CGC, NTSAD Research Associate, praised Meghan, *"Working with Meg during her internship was a true pleasure. She brought expertise and enthusiasm, always volunteering to take on new projects, even on short notice, and showing her genuine care for families.* ***Her dedicated work to help build our new virtual biorepository of patient samples as well as translate many materials and resources for Spanish-speaking families is so greatly appreciated!"***

NTSAD is very thankful for Meghan's help and wishes her all the best as she follows her dream to one day be a geneticist and help rare more families.

Gottlieb Scholarship for Rare Siblings

Applications are now open for the Jeffrey Alan Gottlieb and Stanley N. Gottlieb Memorial Scholarship Funds, which provide financial support for college to healthy siblings of children and adults affected by Tay-Sachs, Canavan, GM1, Sandhoff or an allied disease.

In May 2005, Judy Gottlieb established two separate memorial college funds at National Tay-Sachs & Allied Diseases Association to honor her youngest son, Jeffrey Alan Gottlieb, who succumbed to Tay-Sachs in 1975, and her husband, Stanley N. Gottlieb, who passed away in 2001. **For the last 18 years, Judy has generously awarded scholarships to 56 siblings, some of whom have received multiple awards throughout their education.**

It is Judy's intent that the monies be distributed "for use in their college education, i.e., tuition, books, room and board."

Applications may be submitted until **September 8, 2023,** for consideration this year.

Note: If you previously received a Gottlieb scholarship, and you are still in college, you may apply again!

Apply Today!

NTSAD leads the worldwide fight to treat and cure Tay-Sachs, Canavan, GM1, and Sandhoff diseases by driving

Donate

research, forging collaboration, and fostering community. Supporting families is the center of everything we do.

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