NTSAD Community News

Research, Collaboration, and Community

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

February

February 2022

In this Issue

Dear NTSAD Community,

This month NTSAD's Research Director Valerie Greger and I attended the 18th Annual WORLDSymposium in San Diego where we met with many scientific advisors, clinical researchers, and industry partners dedicated to Tay-Sachs, Canavan, GM1, and Sandhoff diseases. We also attended a variety of presentations and updates about basic science, translational research, and clinical trials for lysosomal diseases. The event brought in more than 1,000 attendees, and NTSAD was among the many exhibitors spreading awareness of our programs, services, and research efforts as well as forging deeper collaboration. Special thanks to NTSAD Board Member Jonathan Katz and Acom Healthcare for pro bono design of the banners featured at our booth, helping us to attract many visitors including a recently diagnosed individual seeking connection and support.

WORLD was an opportunity to acknowledge milestones including nine current clinical trials and look toward the future. We celebrated former NTSAD Executive Director Sue Kahn, who was honored with the Patient Advocate Leader (PAL) Award for her 14 years of leading NTSAD, supporting families, and driving research. We learned from NTSAD's Board President Staci Kallish, who presented on "Fabry in Females: More Than Meets the Eye" as part of the satellite symposia. And we gathered for a Late Onset Think Tank meeting made possible by the Katie & Allie Buryk Research Fund. We left the conference with renewed hope for the NTSAD Community and the joy of connecting in person once again!

Next Monday, February 28th, is Rare Disease Day, and I hope you will join us as we raise awareness and share Why We Care for Rare!

Kathy

Kathleen Flynn
Chief Executive Officer

NTSAD Commemorates 65th Anniversary

NTSAD was founded on hope when 65 years ago three couples, The Berkmans, The Dunkells, and The Sussmans, whose young children were affected by Tay-Sachs, came together to support each other, find answers, and began raising money for research. Their legacy of supporting families, investigating the origin of a rare genetic disease, and funding research has created a strong and enduring Community, enabled countless couples to have healthy children, and led to nine current clinical trials for identifying treatments for Tay-Sachs as well as Canavan, GM1 gangliosidosis, and Sandhoff diseases. They along with hundreds of families were told there was no hope. Today, because of them there is hope, and treatments are on the horizon. We remain indebted to them and the hundreds of families who provide support to each other and drive research.

Later this year, we plan to come together in person to mark our 65th anniversary. More details to follow.

Clinical Trial Updates from Industry Partners

Earlier this month, Aspa Therapeutics hosted an interactive webinar about its upcoming investigational gene therapy program for Canavan disease. To learn more, watch a recording of the webinar available at **www.aspatx.com**. For additional information visit **www.treatcanavan.com** or email **clinicaltrials@aspatx.com**.

Lysogene announced that a third patient has been treated with LYS-GM101 in the company's global clinical trial gene therapy program for GM1 gangliosidosis. **Read more.**

Passage Bio recently shared new interim clinical data for patients with GM1 gangliosidosis participating in the Imagine-1 study. **Read more.**

Sio Gene Therapies provided updates on its GM1 and GM2 clinical trials as well as provided corporate and financial updates. **Read the full release.**

NTSAD Reviews Research Grant Applications

Thanks to all the academics and researchers who sent in letters of intent for NTSAD's next round of funding. Pre-applications will be reviewed by NTSAD's scientific committee, and a select group of applicants will be invited to submit full applications for further review and funding consideration. Applicants invited to advance to the next step will be notified in March. Awards will be announced later this spring.

Learn more about the Research Initiative program.

The Altman Family Shares How They Live Full and Filling Lives

Former NTSAD Board member Stewart “Stew” Altman, who has Late Onset Tay-Sachs disease, and his wife Lorrie were featured in *Patient Worthy's* article *Living a Full and Filling Life with Late-Onset Tay-Sachs (LOTS)* in a two-part series. In part one the **Altmans discuss falling in love, building a family, and *“just doing what you need to do”* to have a happy and fulfilling life – even with a rare disease diagnosis.** Thank you to Stew and Lorrie for your advocacy and sharing your story!

Read the article.

Rare Disease Day: February 28, 2022

Rare Disease Day is less than a week away. Support rare families and help the NTSAD Community raise awareness of Tay-Sachs, Canavan, GM1, and Sandhoff diseases in the following ways:

Follow NTSAD on **Facebook, Instagram, Twitter,** and **LinkedIn,** and read, post, and share Rare stories!

Share *WHY You Care for Rare*, by posting a video or photo holding this sign. Right click the sign to save it as a photo, then print it out. Use the hashtag **#ICareForRareWithNTSAD!** Be sure to check out NTSAD’s staff's videos posted on our social media pages throughout the week.

#ShowYourStripes! If you’re Rare or a Rare caregiver, tell us about yourself or your loved one! NTSAD may feature YOU along with other rare individuals in our awareness efforts.

Attend a Rare Disease Day event:

Register for National Institutes of Health (NIH) virtual Rare Disease Day event on Monday, February 28! Cohosted by the National Center for Advancing Translational Sciences and The NIH Clinical Center, RDDNIH will highlight talks on diversity in rare diseases research, the development of personalized treatments, the use of telehealth during the COVID-19 pandemic, and more! **Register now.**

Grab your favorite striped shirt and join **NORD online for a Rare Disease Day community event on February 28!** This one-hour event will be packed with entertainment from a star-studded guest lineup and offer a place to connect and chat with other rare community members and allies. The event will be held on Monday, February 28 from 1:30 to 2:30pm ET! **Register now.**

Collaboration Enables Carrier Screening

Recently the Evan Lee Ungerleider Fund of NTSAD, the NTSAD New York Area Fund, and the Canavan Foundation, provided immediate and extended family members of an affected child or adult with a discount for comprehensive carrier screening services through NTSAD partner JScreen to help people healthily expand their families.

NTSAD Board member Karen Grinzaid, MS, CGC, CCRC, who is Executive Director of JScreen, reported that 48 people participated in the program and received valuable information to make informed decisions regarding their health and for family planning including:

38 people were found positive for at least one of the 226 diseases included in the screening panel
3 people were found to have two mutations for the same disease and are at risk for symptoms of milder conditions
4 people were found to be carriers of Canavan
2 people were found to be carriers of Tay-Sachs
1 person was found to be a carrier of Sandhoff
No one was found to be a carrier of GM1 gangliosidosis

"It's incredible how many people we've been able to help thanks to the support from these generous funds and foundations. We are so grateful for your partnership!" - Karen Grinzaid

#BeSafeBeTested

Mathew Forbes Romer Foundation's 2022 Gala and Honoree

Don't Miss the Mathew Forbes Romer Foundation's 2022 "Sweetness and Laughter" Gala on April 27th, where NTSAD's former Executive Director Sue Kahn will be honored with the MFRF 2022 "See the Light" Award. Congratulations, Sue! Thank you to the Mathew Forbes Romer Foundation for your partnership and support of NTSAD.

Register for the event today.

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

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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