NTSAD Community News

Research, Collaboration, and Community

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

March 2023

In this Issue

NTSAD ANNUAL FAMILY CONFERENCE

Join us for the 45th Annual Family Conference from June 1 to 4, 2023 in Reston, Virginia. In addition to Rare individuals and families, the Conference brings together the entire NTSAD Community, including researchers, patient advocates, and industry members. To date, more than 40 families and more than 100 people have registered! All are welcome!

If you haven't already, please register to attend now.

Reserve your hotel room.

You also can check out the current schedule for the Conference. And remember to check back often, as we will be adding more details as we get closer to the Conference dates.

For affected adults and families in need of financial assistance, Helping Hand grants are available. **Apply here.**

Thanks to the generosity of our 2023 Conference sponsors who make it possible for affected children and adults and their families to experience four days of connection, community, and support.

2023 Annual Family Conference Sponsors

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Learn more about sponsor opportunities. Please confirm your sponsorship no later than April 15th to be included in Conference materials. Questions? Email Director of Development and Communications, Susan Keliher.

Donate to the Conference

Passage Bio GM1 Gangliosidosis Clinical Trial Update

Passage Bio recently shared encouraging news about its GM1 Gangliosidosis study. **Passage Bio's drug, PBGM01, continued to demonstrate a biological effect in the first six patients in its Imagine-1 study.** Passage Bio also announced planned expansion of Imagine-1 study to treat patients with gene therapy at a higher dose and expect to dose the first patient at this level in the second half of 2023.

Learn more.

Advancing Research through Natural History Studies

Understanding the progression of a rare disease is critical to the development of a clinical trial. As a Rare individual or family, there are many ways to contribute to research through sharing your experiences, including through participation in a natural history study. **Currently there are open natural history studies for Canavan, GM1, and GM2 (Tay-Sachs and Sandhoff) diseases.**

If you are considering participating, we encourage you to contact NTSAD's Family Services team to discuss the benefits and risks.

Learn more about current natural history studies.

Contact the Family Services team.

NTSAD and Buryk Research Fund Host LOTSS Meeting to Advance Research

On March 7th, NTSAD and the Katie and Allie Buryk Research Fund convened researchers, industry members, and key stakeholders to discuss the latest in research, foster innovation, and advance treatments for Late Onset Tay-Sachs and Sandhoff (LOTSS) diseases.

Keynote speaker, Krystof Bankiewicz, MD, PhD, discussed the development of gene therapy for two conditions, AADC Deficiency and Parkinson's disease, sharing how gene therapies for these conditions may

lead to discoveries and assist in the development of clinical trials for LOTSS including where in the body treatments should be delivered.

Researchers provided updates on the following promising research:

The development of AAV (adeno-associated virus) gene therapy in a sheep model for Tay-Sachs and the development of biomarkers to assess if the gene therapy is working. Thus far, the data indicates gene therapy is improving neurological symptoms in the sheep.

Work on base editing, a technique used to directly modify the DNA sequence was discussed. **Progress is being made to develop this editing system in a mouse model to correct a DNA change common in many individuals with LOTSS.**

In addition, there was much discussion on forward-leaning efforts toward clinical trial design and outcome measures should positive data continue, such as the use of artificial intelligence tools in studying and treating neurogenetic conditions. Also, there was a presentation on a broad survey of the LOTSS Community's attitudes about their symptoms and therapies to inform the development of clinical trials.

Thank you to **The Buryk family, Dr. Bankiewicz**, and presenters: **Xuntian Jian, PhD; Rick Proia, PhD; Cristina Sampaio, MD, PhD; Swati Sathe, MD, MS; Ben Solomon, MD; Toloo Taghian, PhD;** and **Camilo Toro, MD**, and everyone who contributed to Late Onset Tay-Sachs and Sandhoff (LOTSS) Diseases: Updates for Science and Industry meeting and everyone working towards finding treatments for the LOTSS Community.

NTSAD's New Research Associate Caroline Aragón

NTSAD welcomes Caroline Aragón MS, CGC, as our first ever Research Associate! **A certified genetic counselor, Caroline is deeply committed to addressing rare diseases and has extensive clinical training.** In addition, she has conducted “hands-on” genetic testing, coordinated clinical studies, and performed basic science laboratory research. In her role as Research Associate, Caroline will help drive research and manage an international network of researchers, clinicians, and industry partners who collaborate with NTSAD through the Newborn Screening Consortium, LOTSS Think Tank, Research Committee and Research Initiative grant process, and the Scientific Advisory Council.

To reach Caroline, email her at **caragon@ntsad.org**.

Feeling Green? Get Screened!

If you plan to celebrate your Irish heritage this St. Patrick's Day, consider sharing with your family and community the importance of genetic testing. **It is believed that one in 50 individuals of Irish ancestry are carriers of Tay-Sachs disease.**

For people planning a family, it is important to know if you are a carrier of any rare genetic diseases. When both parents are carriers of Tay-Sachs, Canavan, GM1, or Sandhoff, each child has a 25% chance of having the disease. The carrier rate for the general population for Tay-Sachs and GM1 diseases is 1/250. Like the Irish, people of British Isle, French Canadian, Louisiana Cajun, Ashkenazi Jewish descent are also considered to be at higher risk. For Canavan disease, the carrier rate for the general population is 1/300. People of Ashkenazi Jewish descent are at high risk with a carrier rate of 1/55. For

Sandhoff disease, the carrier rate is approximately 1/600 in the general population. **Learn more about genetic testing.**

Nowadays, getting screened is as easy as speaking to a genetic counselor and having a testing kit sent to your home. **Empower your family by getting tested.**

Check out the Jewish Genetic Disease Consortium website.

Please contact NTSAD at **info@ntsad.org** with any specific questions about genetic testing.

NTSAD Community Raises Rare Awareness

"We want the world to know that even though we are a Rare family, we are here. We live in a world full of isolation and routine. It's very lonely. If you know someone Rare, remember to check on them often. Offer your help. Offer a meal. A shoulder to cry on. Go over and visit. Make the time. It means a lot to those families. Rare is real." - Joshua and Cynthia, Dad & Mom to Levi

Throughout last month leading up to Rare Disease Day on February 28th, the NTSAD Community came together and honored Rare loved ones by showing the world that Rare is Real! Thank you to all the families who shared with us what they want the world to know about Rare. You can read all their answers and see the faces of beautiful, Rare children and adults on the **NTSAD Facebook page**.

NTSAD thanks the entire Rare Community for the tireless efforts to advocate, raise awareness, and fund research on Rare Disease Day, throughout Rare Disease Month, and all year long. Together, we work to find effective treatments and support Rare Families.

Leading up to Rare Disease Day, NTSAD CEO Kathy Flynn, Research Director Valerie Greger, and Director of Family Services, Diana Jussila, attended the 19th Annual WORLD Symposium in Orlando Florida and met with researchers and members of industry from around the globe developing potential therapies for lysosomal diseases.

Although Rare Disease Month is behind us, you can be an advocate for Rare all year long! Check out **NTSAD website** for resources and follow us on social media (linked below) so you never miss an opportunity to spread awareness.

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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