NTSAD Community News

Research, Collaboration, and Community

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

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NTSAD ANNUAL FAMILY CONFERENCE

2023 Registration Now Open!

Join us in Reston, Virginia, for NTSAD's 45th Annual Family Conference from June 1 to 4, 2023. All are welcome— families, clinicians, researchers, industry members, and rare allies.

The Annual Family Conference brings together families and individuals coping with a diagnosis of Tay-Sachs, Canavan, GM1, or Sandhoff disease to receive support, connection, and community. The four-day event offers sessions for affected adults, parents and partners providing care, and families experiencing grief and loss. On Friday, June 3, researchers and clinicians share the updates on the latest research, current projects, and clinical trials. During a Commemoration Ceremony we gather to honor and remember lost loved ones. There are activities and camps for affected and healthy children throughout the Conference as well as opportunities to connect for all.

Register today!

Helping Hand Grants are available for families and individuals who need financial support.

Helping Hand Grant Application

The Annual Family Conference is made possible through the generosity of our Sponsors. **Learn more about the conference sponsorship opportunities.** For more information, contact Susan Keliher, Director of

Development and Communications
at **skeliher@ntsad.org.**

A Rare Mom's Goals for Her Child

"What are your goals for your daughter?"

It's a question I get asked all the time. By the social worker and her complex care coordinator. By all three physical therapists (PT's) that she sees at three, separate locations, the speech therapists, and occupational therapists, too.

It was a hard question for me to answer, especially at first. I had difficulty comprehending what her future would look like. Goals? She has a degenerative disease, don't things just get worse from here?

The answer we had then is the same as the one we have now that she is in a clinical trial. **We want our two-year-old daughter to have as much autonomy as possible and the ability to make her own choices.**

The ability to make a choice is a big deal. When she learned to roll, she no longer was stuck playing with whatever toy I gave her, as long as it is only a roll away! When presenting her with blueberries and strawberries and letting her reach for them, I learned that she will always pick strawberries, although she will eat blueberries when that is the only option. Being able to indicate which foods you truly enjoy is a big deal as well. And clothes. It turns out that she has an opinion about her clothes, too.

This week, I went to an equipment clinic with my daughter where we ordered some new equipment for her, including a wheelchair for when she goes to preschool this fall. I spent a lot of time with the two PT's and the equipment sales rep talking about which chair would be best for our daughter given her evolving needs.

"You want to give her the ability to be as much like the other kids as possible. To go where she wants, when she wants," the rep said. So I picked a chair that she will (I hope) be able to learn to control on her own, to wheel herself around her classroom, or go where she wants when we are at the zoo or museum, rather than wherever I push her stroller.

And then it struck me! Those of us who are able-bodied see the wheelchair as a symbol of all the things people can't do. One we use as we heal from an injury or when we get old, and our bodies can no longer do what they once did. **But for my child, a wheelchair, especially one that she can control, will bring her so much autonomy and the ability to make her own choices.** Yes, it is different for her than for the other kids, but it is also a very wonderful thing that will give my girl the opportunity to "run" away and hide from me in a store like all kids do.

So here's to mobility devices! The ones that allow us to take our kids out of the house in safe, comfortable seating, so they can feel the sun on their skin and spend time with their family or friends, even if they can't control those devices on their own.

And here's to my beautiful daughter gaining more autonomy and the ability to make new choices, including getting into a little trouble.

Rare Disease Day: February 28, 2023

There are more than 7,000 rare diseases affecting more than 400 million people worldwide. **February 28 is Rare Disease Day,** a global movement that highlights the need for equity in social opportunity, healthcare, and access to diagnosis and therapies for people living with a rare disease. Join us in raising awareness of Tay-Sachs, Canavan, GM1, and Sandhoff diseases next month. ***Rare is Real,* and You and Your Support Make a Real Difference!**

Here are three ways you can participate and show your support!

Follow NTSAD on **Facebook,** **Instagram,** **Twitter,** and **LinkedIn,** and read, share, and join our **#RareIsReal** movement!

Join the National Institute of Health for its annual Rare Disease Day virtual event on February 28, 2023, from 9 am to 5 pm Eastern Time! **Check out the speakers and more.**

Purchase a "Rare Is Real" sweatshirt and other fun items, so you can **#ShowYourStripes,** spreading Rare awareness and raising funds for NTSAD's Research Initiative. **Check out all the items available here.**

Remembering Fred Horak and the Discovery of Jacob Sheep

Fred Horak, who along with his wife, Joan played an integral role in advancing research for identifying treatments for Tay-Sachs diseases. Sadly, Fred passed away this month, and we would like to honor him by sharing the Horaks' incredible contributions to the NTSAD Community.

In 1999, Texas farmers Fred and Joan Horak noticed two lambs among their flock of Jacob's sheep had developed signs of a nervous system disorder. Fred and Joan consulted a veterinarian and realized that the sheep had an inherited disease. They kept good records on their flock and knew the parental origin of the affected sheep. They could have removed these animals from their breeding stock, but instead continued to care for the affected sheep and breed the sheep, so that perhaps someday they would be better understood the gene that caused the disease which would ultimately serve a greater purpose.

It took more than a decade to solve the mystery. Brian Porter, DVM, a veterinary pathologist at Texas A&M College of Veterinary Medicine & Biomedical Sciences, referred the Horaks to Edwin Kolodny, MD, at New York University Medical Center. Dr. Kolodny's lab discovered that the Jacob sheep had Tay-Sachs disease. With funding from NTSAD and the Horak's donation of the affected Jacob sheep, the veterinary school at Auburn University was able to conduct a large animal gene therapy study, which paved the way for clinical trials in humans.

In 2010, Fred and Joan were honored with NTSAD's Above and Beyond Award, recognizing individuals for the significant impact they have made on the NTSAD Community. Here is an excerpt of the award nomination from rare disease pioneer Edwin Kolodny, MD.

"The many hours the Horaks spent preparing lists of their flock for breeding, in the fields and pens on their farm caring for their animals, encouraging other breeders to test their flocks of Jacob sheep, telephoning and emailing me and our Gene Therapy Consortium colleagues, and sending blood spots and tissue specimens to us, are too numerous to count. This is a couple who have seized an opportunity thrust upon them to make a better world for all, for the sheep that they love, but more so for the greater good of humanity. They have set a very high standard in humanitarian service."

The NTSAD Community remains grateful to Fred and Joan Horak for their tireless efforts, which led to groundbreaking research and clinical trials for Tay-Sachs disease.

Fred and Joan Horak - Above and Beyond

Fred and Joan Horak were recognized at NTSAD's Annual Family Conference with the Above and Beyond Award in 2010.

Pictured left to right: Dr. Kolodny, Fred Horak, and Marion Yanovsky (NTSAD New York Area Fund).

NTSAD's Research Initiative

NTSAD's Research Initiative's recent request for proposals yielded 14 first-round applications from researchers seeking for funding during the 2023 grant cycle. Applications span all four diseases: Tay-Sachs, Canavan, GM1 gangliosidosis, and Sandhoff.

Following review by the Research Committee and NTSAD's Scientific Advisory Council, and with input from the newly relaunched Corporate Advisory Council, NTSAD will invite projects with the most scientific merit to submit full proposals. The Research Committee will then review proposals and make recommendations for funding while considering the needs of the community and alignment with NTSAD's mission. Funding decisions will be announced in June 2023.

NTSAD Board Seeking Member with Finance Background

Seeking community support! The NTSAD Board is seeking a board member for the 2023 slate with experience in finance and/or accounting. Do you have a relative or family friend who has a finance background who may be interested in giving back to the NTSAD Community? Our goal is to elect a finance-savvy board member who has connections to one of our families as part of our 2023 board slate. If you or someone in your network fits the bill, please reach out to Kathy at **kflynn@ntsad.org** as we would welcome a chance to connect!

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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