NTSAD Community News

Research, Collaboration, and Community

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

March 2022

In this Issue

Dear NTSAD Community,

This March marks two years since COVID-19 drastically changed our world. As we usher in spring, COVID cases continue to decline, there is renewed hope as a bit of normalcy returns to our lives, and clinical trials for our Community continue to show promising early data. It is our hope that will join us in Denver this July for NTSAD's 44th Annual Family Conference when we plan to gather in person once again to connect and provide support to families, and to hear the latest updates in research and clinical trials from industry members.

March also brings St. Patrick's Day, a celebration for people of Irish heritage including my family and millions who are part of the Irish diaspora. This year I approach the festivities with the knowledge that those of Irish and British Isles decent are at greater risk of being a carrier of Tay-Sachs or GM1. It is believed that one in 50 individuals of Irish ancestry are carriers. I have encouraged my loved ones to partake in genetic testing and carrier screening to help protect future generations. **It is important to note that anyone can be a carrier of Tay-Sachs, Canavan, GM1, and Sandhoff diseases. We encourage everyone across all ethnicities to participate in genetic screening.**

I invite you to spread awareness and help prevent the anguish of having a child diagnosed with a debilitating or terminal disease through education about carrier screening. Further below in this newsletter you can learn more about carrier screening, including resources.

This month we also continue to commemorate NTSAD's 65th Anniversary and remember the pioneers and many other dedicated clinicians and researchers whose discoveries laid the groundwork for awareness, diagnosis, prevention, and progress toward finding a cure. We thank you and acknowledge you for all you do to support our rare families.

Sincerely,

Kathleen Flynn
Chief Executive Officer

Support Families and Sponsor the Annual Family Conference

NTSAD's 44th Annual Family Conference is just four months away! **For the first time in more than three years, NTSAD families, researchers, clinicians, and rare allies will be gathering in person from July 7 to 10 in Denver, Colorado!**

The goal of the conference is to provide families and individuals affected by Tay-Sachs, Canavan, GM1, and Sandhoff diseases coping with a diagnosis, providing care, or healing from a recent or enduring loss with the latest updates in research, resources, and support.

"I look forward to attending the NTSAD conference. Having a child affected by these terrible diseases is an isolating experience. Being with other families, having my children be with other kids who have lost a sibling in this manner, is really only available to us when we come. It is a comforting balm to be with everyone and to feel the specialness of all your children, to be in company as we grieve at whatever point we are in this journey that is not confined to the life of our affected children. I look forward to any opportunity that helps me feel closer to my Lucy, and anything related to NTSAD does that for me." - **Emily Day, Mom to Lucy, Infantile GM1**

Help us make the conference possible and affordable for families by becoming a 2022 Conference sponsor. **Learn more about the conference sponsorship opportunities.**

Should you wish to sponsor the Conference contact Susan Keliher, Director of Development and Communications at skeliher@ntsad.org by April 15, 2022 to be included on conference materials.

If you're an affected family interested in attending your first in-person conference, Family Services Manager Becky Benson is holding an informational Zoom meeting on **Tuesday, March 22, 2022 at 5 pm EDT. Email becky@ntsad.org to receive the link to join. In addition, for families who need financial assistance email about becky@ntsad.org Helping Hand Grants.**

Canavan Gene Therapy Receives Fast Track Designation

Myrtelle, Inc. received the U.S. Food and Drug Administration's (FDA) fast track, rare pediatric disease, and orphan drug designations for the company's proprietary gene therapy for the treatment of Canavan disease.

"The FDA's decision to grant these designations for our investigational gene therapy utilizing rAAV-Olig001-ASPA aligns with our mission to provide treatments for patients where few, if any, options exist and highlights the urgency of developing a treatment for patients with Canavan disease, a devastating disease of young children which results in short life expectancy," said Nancy Barone Kribbs, PhD, Senior Vice President of Regulatory Affairs at Myrtelle.

Read press release.

Advocate for STAT Act - Speeding Therapies Today

In one minute, you can help speed up the development process for a rare disease drug, which currently takes and average of 15 years and affects more than 30 million Americans with a rare disease. Support the STAT Act — Speeding Therapy Access Today Act of 2021, H.R. 1730/S. 670.

The STAT Act is a bipartisan bill that was created with the input of the rare disease community aimed at improving the development of and access to therapies for the rare disease community. The centerpiece of the STAT Act is the creation of a Rare Disease Center of Excellence at the US Food and Drug Administration. The STAT Act will:

- Accelerate rare disease therapy development
- Optimize interagency coordination,
- Advance science-based regulatory policies, and
- Facilitate access to therapies.

Thanks to The EveryLife Foundation and so many rare families and allies meeting with their representatives in Congress during Rare Disease Week on Capitol Hill, the Speeding Therapy Access Today (STAT) Act will be discussed in a Congressional hearing on Thursday, March 17th.

The STAT Act's inclusion in the Energy & Commerce Committee hearing marks a major milestone in passing targeted and impactful policy reforms at the Food and Drug Administration (FDA). **Please take a minute to ask your member of Congress to co-sponsor the STAT Act today before March 17th!**

Learn more about the STAT ACT and find your representative in Congress.

TORCH Award Nominations for Rare Advocates

Do you know someone inspiring with their rare advocacy? Nominate them for a Torch Light Award. Sanofi, formerly Sanofi Genzyme, announced it is accepting nominations for the TORCH Light Awards through April 15, 2022.

The annual program recognizes individuals who have made a meaningful contribution to a lysosomal storage disorder community, or to a disease area within Sanofi's rare disease research and development program. Individuals receiving TORCH Awards have found a way, in their own capacity, to bring awareness to rare diseases and to educate, empower, advance, or connect patients. NTSAD Parents Brian and Sherri Manning were honored with a TORCH Award for their rare advocacy in memory of their son Dylan and were featured in last year's show. You can be, too. Nominate yourself or someone you know and save the date for this year's show on Thursday, August 25, 2022.

Read more about the nomination process.

Watch the 2021 show online!

Remembering Pioneers

It is often said that at NTSAD we stand on the shoulders of all the families and pioneers who came before us. In last month's issue we highlighted NTSAD's founding families the Berkmans, Dunkells, and Sussmans. This month, we look at the researchers and clinicians whose work has led us to where we are today with multiple active clinical trials for Tay-Sachs, Canavan, GM1, and Sandhoff diseases.

In 1881, Warren Tay, a British ophthalmologist, described a patient who had a cherry-red spot on the retina of the eye, a symptom of infantile Tay-Sachs disease and often the first indicator for families seeking a diagnosis. Several years later, Bernard Sachs, a New York neurologist whose work provided the first description of the cellular changes occurring in patients with Tay-Sachs disease. He noticed the familial nature of the disorder and observed numerous cases. He noted that most babies with Tay-Sachs disease at that time were of Eastern European Jewish origin. Today, we know Tay-Sachs affects people of every ethnicity and background.

In August 1969, Shintaro Okada MD and John S. O'Brien MD published their discovery of the Hexosaminidase A deficiency in Tay-Sachs patients. Their discovery led to the first community screening for Tay-Sachs in May 1971 in Bethesda, Maryland. The specific gene that carries Tay-Sachs was identified in the late 1980s, and by the mid-1990s more than 75 different mutations had been identified. Currently there are more than 100 mutations reported across all ethnic groups.

In 1965, Horst Jatzkewitz, Hartmut Pilz, and Konrad Sandhoff first noticed what later would be named Sandhoff disease in Germany. Initially these men were studying the biochemistry of different enzymes and found an exceptional case of Tay-Sachs. These doctors classified Sandhoff as an abnormal Tay-Sachs disease and published their findings in the *Journal of Neurochemistry*. In 1968, Sandhoff, along with U. Andreae and Jatzkewitz further discussed the disease in the journal *Life Sciences*.

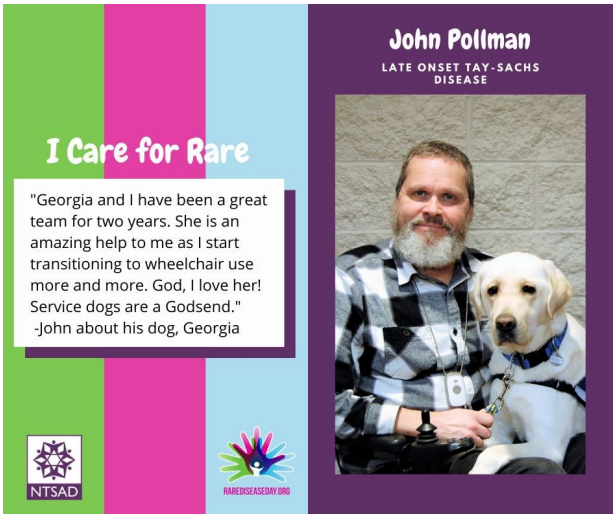
In 1931, Myrtelle May Canavan MD, one of the first female pathologists, published a paper about a disease that had claimed the life of a 16-month-old baby. Canavan noted that the child's brain had a soft, spongey section that had turned white. She was the first to describe this disease which would become known as Canavan disease. In 1993, Rueben Matalon MD discovered the gene that causes Canavan from tissues provided by several Canavan families. This discovery led to carrier screening and prenatal testing for the disease.

John Caffey MD likely gave the earliest description of GM1 in 1951 while discussing prenatal onset of Hunter-Hurler disease. Dr. Landing gave the first definitive description of Gangliosidosis-1 (GM1) in 1964, which had previously been called "Hurler variant," "pseudo-Hurler disease," and "Tay-Sachs with visceral involvement."

These scientists were critical to identifying symptoms and causes so that doctors could make a diagnosis for families and scientists could develop prevention efforts. These discoveries paved the way for today's pioneers and clinicians like Florian Eichler MD, Guangping Gao PhD, Heather Gray-Edwards DVM, PhD, Michael Kaback MD, Edwin Kolodny MD, Douglas Martin PhD, Miguel Sena-Esteves PhD, Cyndi Tifft MD PhD, and many others. Their tireless devotion and important life's work have propelled potential treatments and therapies for Tay-Sachs, Canavan, GM1 and Sandhoff diseases.

NTSAD Families Spread Awareness on Rare Disease Day

Thank you to all the families and individuals who shared their rare, loved ones and raised awareness for Tay-Sachs, Canavan, GM1, and Sandhoff diseases on Rare Disease Day in February 28 and throughout the month of February. If you haven't already, check out NTSAD's Care for Rare posts on our **Facebook** page featuring nearly 50 rare individuals and what makes them special.



Prevention and the Importance of Carrier Screening

For people planning a family it is important to know if you are at risk for being a carrier of a rare genetic disease. When both parents are carriers of Tay-Sachs, Canavan, GM1, or Sandhoff, each child has a 25% of having the disease. The carrier rate for the general population for Tay-Sachs and

GM1 diseases is 1/250. French Canadians, Louisiana Cajuns, and Ashkenazi Jews are all considered to be at high risk with a carrier rate of 1/27. However, individuals of any family origin can also be carriers.

For Canavan disease the carrier rate for the general population is 1/300. Ashkenazi Jews 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, and it is not yet clear whether Sandhoff disease is more common in any particular population, but it may have a higher carrier rate in several, somewhat isolated populations. This month begin a conversation about the importance of carrier screening with your family, your healthcare provider, and in your community.

Learn more about carrier screening.

Talk to a genetic counselor and get tested.

Check out the Jewish Genetic Disease Consortium website.

Please contact NTSAD at **info@ntsad.org** with any specific questions about carrier screening.

Tay-Sachs and Irish Heritage

Are you celebrating your heritage this St. Patrick's Day? Did you know individuals with Irish ancestry are believed to be at higher risk than the general population for Tay-Sachs carrier status?

NTSAD parents Aaron and Kathryn Harney, who are of Irish decent, continue to spread awareness in honor of their late son Nathan, who was diagnosed with juvenile Tay-Sachs and passed away at four. Years ago, the family promoted a study to further identify carriers and the incidence rate among people of Irish heritage. Help continue Nathan's legacy and get tested.

Watch their story below.

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

STAFF
Kathleen M. Flynn, CEO
Becky Benson, Family Services Manager
Sydnie Dimond, Development and Communications Associate
Valerie Greger, Director of Research
Susan Keliher, Director of Development and Communications
Diana Pangonis, Director of Family Services

NTSAD
2001 Beacon Street
Suite 204
Boston, MA 02135