NTSAD Community News

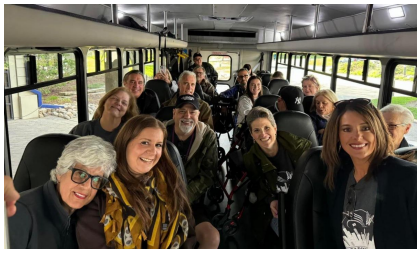
Research, Collaboration, and Community

June

June 2024

In this Issue

Late Onset Natural History Study Published!



To date, no validated clinical outcome assessments (COA) for Late Onset GM2 gangliosidoses (Tay-Sachs and Sandhoff diseases) exist, but a recently published natural history study following 23 individuals with Late Onset GM2 for five years has been published. The study followed clinical changes in neurological, functional, and quality of life- to inform the design of future clinical interventional trials. Researchers found slow but significant worsening of symptoms and disease burden.

"This study is a great example of what patient organizations can do to become ready for clinical trials." - Gerald Cox, MD PhD, NTSAD Board Member and Chair of NTSAD's Research Committee

Individuals affected with Late Onset GM2 who attended NTSAD's Annual Family Conferences between 2015 and 2019 underwent annual clinical outcome assessments, which contributed to this natural history study.

"This LOTSS natural history project involved the dedicated efforts of many key experts in the GM2 space, many of whom are affiliated with NTSAD. A special thanks to all those who contributed to this study — especially the adults with late onset GM2 who participated in the assessments, members of the scientific community, and Diana Jussila, NTSAD's Director of Family Services." — Kathy Flynn, CEO NTSAD

Read the paper.

FDA Taps Myrtelle for Pilot Program

The U.S. Food and Drug Administration (FDA) has selected Myrtelle's rAAV-Olig001-ASPA gene therapy candidate for the treatment of Canavan disease for inclusion in

the agency's Support for Clinical Trials Advancing Rare Disease Therapeutics (START) pilot program.

The START pilot program aims to accelerate the development of novel drug and biological products for rare diseases. rAAV-Olig001-ASPA was selected as one of a few CBER-regulated products based on eligibility criteria, including clinical benefit for rare diseases with unmet medical needs and the sponsor's ability to accelerate development to market application. The program facilitates ad hoc communication with FDA staff to help sponsors achieve regulatory milestones and expedite product development.

The objective of the program is to accelerate the development of gene therapies for rare diseases that lead to significant disability or death within the first decade of life by facilitating more frequent advice and regular communication with FDA staff. Myrtelle is among the six participants in this new FDA initiative.

*"**Acceptance into the START pilot program is an honor in that it recognizes rAAV-Olig001-ASPA as a candidate for accelerated development as a potential treatment for Canavan disease.** Opening the lines of communications beyond traditional meeting pathways provides the opportunity to quickly address development issues that would otherwise delay progression to market application. **We are encouraged by the opportunity to facilitate the development of a potential treatment for Canavan children who are without treatment options,"*** said Nancy Barone Kribbs, PhD, Senior Vice President of Global Regulatory Affairs at Myrtelle.

Read the press release.

Assay Developed for Newborn Screening for Canavan Disease

A recently published manuscript describing the development of a new assay method for newborn screening for Canavan disease could enable earlier diagnosis. The pilot study, **Quantification of N-acetyl-l-aspartate in dried blood spots: A simple and fast LC-MS/MS neonatal screening method for the diagnosis of Canavan disease** was performed on 25 people with Canavan disease. Given that promising treatments may be on the horizon, early diagnosis of Canavan disease is crucial for effective treatment.

Authors include Christian Posern, Benjamin Dreyer, Sarah L Maier, Florian Eichler, Michael H Gelb, René Santer, Annette Bley, and Simona Murko. NTSAD has had a long history of collaboration with several of the study's authors.

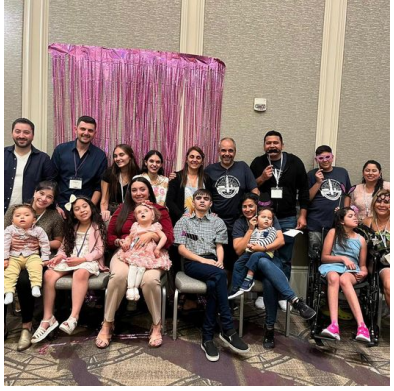
Read the abstract.

Read the full article.

Research Updates Now Available in Spanish

NTSAD's Annual Family Conference includes an entire day dedicated to topics related to research, clinical studies, and updates in the field of each disease NTSAD supports.

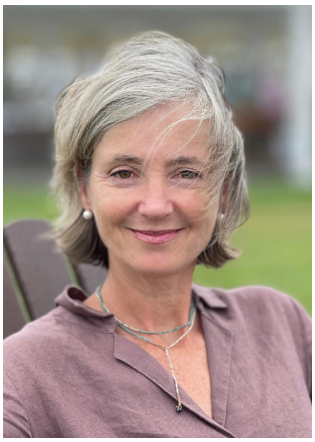
Summaries of each breakout session for Canavan, infantile and juvenile GM1, Tay-Sachs and Sandhoff, and Late Onset Tay-Sachs



and Sandhoff are available in English- and Spanish on the NTSAD website.

Read Conference research summaries in Spanish and English.

Thank You, Blyth Lord, for Your Years of Service



Blyth Lord, Mom to Cameron, Eliza and Taylor and Founder of Courageous Parents Network (CPN), wraps up more than two decades of service on the NTSAD Board, including serving as Board President. Blyth, a visionary, lended her voice and experiences through the years as the community witnessed the advent of clinical trials, the development of a five-year strategic plan for NTSAD, and the overall growth of the organization.

Blyth's second daughter, Cameron, was diagnosed with Tay-Sachs disease at the age of six months. In addition, Blyth's nephew Hayden —Cameron's first cousin—was also diagnosed with Tay-Sachs disease, one month before Cameron's diagnosis, when he was 18 months old. Hayden and Cameron's fathers Charlie and Tim Lord are identical twins who both married carriers—Blyth and Alison who were, also, best friends in college. The twins' genetic variants had never been seen before, and the likelihood of these two brothers marrying carriers and having affected children was 1 in 80,000,000. Both Cameron and Hayden died before the age of three.

After their profound losses, Blyth, Charlie, Tim, and Alison founded the Cameron and Hayden Lord Foundation to raise awareness and more than one million dollars in funds to help NTSAD support families like theirs and advance research. Blyth did not stop there. She founded CPN in 2013 to provide education, community and advocacy as well as information, skills, tools, and support for parents navigating their child's illness.

Blyth's Fellow NTSAD Board Members Shared:

"Blyth's dedication to NTSAD and our families is unparalleled. *We can never thank her enough for her mentorship, guidance, leadership, and love during her service on the board, and during the past 25 years. We will miss her voice and ideas at the Board level, but know she will never be far from NTSAD."* — Bonnie Davis, Mom of Adam and NTSAD Board Member

"Blyth Lord embodies power, grace, and compassion. ***To be able to have accomplished all that she has, both for NTSAD and Courageous Parents Network, is a feat that few could have matched,*** *and we are all better for having Blyth in our lives."* — Jamie Ring, NTSAD Board Member

"Blyth had been a wonderful role model for what a thoroughly engaged NTSAD board member should be. ***Thank you, Blyth, for showing us the way and for all you have done to strengthen this organization."*** — Martha Kleinman, NTSAD Board Member

BridgeBio Gene Therapy Celebrates the Americone Dream

Here's the best reason to eat ice cream. Proceeds from Ben & Jerry's Americone Dream flavor created for Stephen Colbert, host of The Late Show, are allocated to charity. NTSAD has been chosen as a beneficiary — thanks to a connection with an NTSAD family!

Thank you to BridgeBio, parent company of Aspa Therapeutics, for organizing an Ice Cream Social in support of NTSAD, featuring the scrumptious Ben & Jerry's Americone Dream!

"Supporting families is the true purpose of our work and our partnership with NTSAD allows us to deepen our commitment of service to the community. An activity like this gives the entire team at BridgeBio Gene Therapy the opportunity to show our support!" — *BridgeBio Gene Therapy Chief Scientific Officer, Clayton Beard, PhD.*

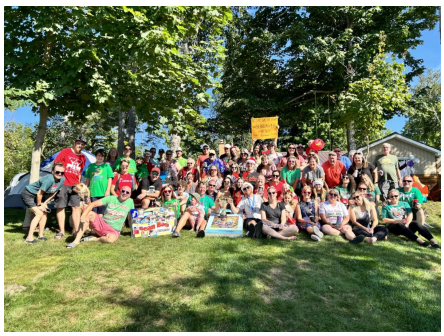
You can help families, too! Pick up a pint of *Ben & Jerry's Americone Dream* today. Enjoy while watching *The Late Show with Stephen Colbert.*

Meanwhile...if you have a connection to a business or foundation that can help raise awareness or support families by donating, please contact NTSAD's Director of Development and Communications, Susan Keliher **here** or call the office at 617-277-4463.

Day of Hope 2024 - Host Your Own Event

Each year, the NTSAD Community unites to raise awareness and funds for Tay-Sachs, Canavan, GM1, and Sandhoff diseases by hosting a Day of Hope event.

From June to October each year, rare families host walks, poker runs, cornhole tournaments, trivia nights, golf tournaments, Facebook fundraisers, t-shirt campaigns, and more in support of NTSAD families and research.



You too can hold your own Day of Hope event in your community.

To learn more about **Day of Hope**, plan your own event, and/or raise money for **Day of Hope**, contact Becky Benson, NTSAD's Family Services Manager **here** or call Susan Keliher, NTSAD's Director of Development at 617-277-4463 or email her **here.**

Mark Your Calendars - *Imagine & Believe* on Thursday, November 7th

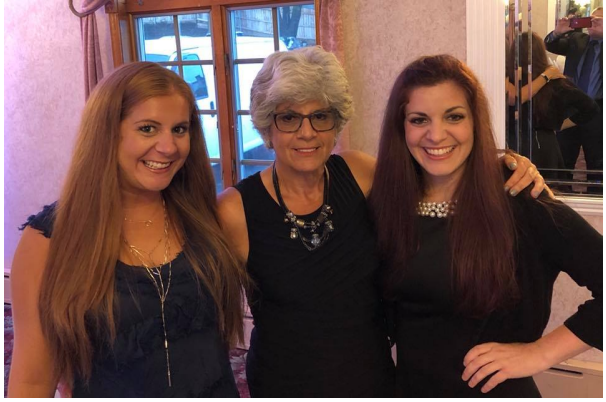
Save the date for NTSAD's signature fundraising event, ***Imagine & Believe,* at the Le Méridien Cambridge on November 7th from 6-9 pm ET!**

This year we honor the Buryk Family for their advocacy in raising awareness and driving research forward with their incredible efforts to raise funds leading to over one million dollars!

Join the community, fellow supporters and those

who seek to make a difference on November 7th.

There will be a reception, silent auction, and inspiring speaking program as we raise critical funds for family services and programs, advocacy, and research. **Buy your tickets today!** Sponsorship opportunities are available. **Learn more here.**



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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