NTSAD Community News

Research, Collaboration, and Community

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

July

July 2023

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Azafaros Recruiting for GM2 Clinical Trial

Azafaros, a biotech company based in the Netherlands, began recruiting for its RAINBOW Study, a phase 2 multinational trial for Tay-Sachs, Sandhoff, (known collectively as GM2) and Niemann-Pick Type C. The study will be held in Brazil and the United States.

Recently, NTSAD and the National Niemann-Pick Disease Foundation hosted a community meeting with Azafaros about the RAINBOW study and answered questions from rare families. The Phase 2 RAINBOW study is a randomized, double-blind, placebo-controlled, multicenter, 12-week trial assessing the safety, tolerability, pharmacokinetics, and pharmacodynamics of AZ-3102 in patients with GM2 gangliosidosis and NP-C. The study will enroll 12 patients in total (six with GM2 and six with NP-C), aged 12 to 20 years, each of whom will receive either the low dose, high dose, or a placebo. Patients who complete the 12-week study period will be offered a double-blind extension if approved by the country's health authorities. The aim of the short study is to determine the clearance of AZ-3102 from the body and the effect of two different doses in patients to identify the target dose for Azafaros' planned Phase 3 pivotal studies.

Watch the community meeting with Azafaros about the RAINBOW Study, now available on NTSAD's YouTube channel.

NTSAD's New Research Directory of Patient Samples

This July, NTSAD launched a first of its kind inventory of patient samples from children and adults affected with Tay-Sachs, Canavan, GM1, and Sandhoff diseases to advance research and accelerate treatments. NTSAD is known for forging collaboration and toward that goal, NTSAD has created a virtual biorepository to facilitate the sharing of research samples including cell lines and patient samples among researchers and clinicians. NTSAD serves as a connection point between researchers looking to locate and share samples, but will not physically collect or house biological samples.

If you are a researcher either seeking samples or storing samples, **visit our website for more information.**

Questions? Contact Research Associate Caroline Aragón, MS, CGC at **caragon@ntsad.org.**

Working for Noa – Canavan Gene Therapy

Lee and Lori Greenwood, whose daughter Noa has Canavan disease, recently visited with researchers, Guangping Gao, PhD, and Dominic Gessler, MD, PhD, at the University of Massachusetts Chan Medical School (UMCMS) to say thank you and share Noa's progress after participating in the researchers' Canavan gene therapy trial licensed by Aspa Therapeutics.

Diagnosed at 11 months, the Greenwoods felt lucky that Noa was eligible for the CANaspire trial. Noa became the third child in the study to receive a dose of gene therapy.

"*I can't believe it. I'm shocked. Looking at her, she is not a two-year-old patient with Canavan*," said Elisabet Mandon, PhD, associate professor of microbiology & physiological systems and part of the gene therapy research team.

"*What Noa is doing is just what my grandchildren would do*," said Dr. Gao. "*Canavan research is part of my life, and to see gene therapy make a difference, it's just so satisfying."*

Meeting a child who received gene therapy also guides researchers. ***"In clinical practice, you identify problems or diseases, and the treatment options are limited. But bringing those questions back to the lab and trying to find answers, and then seeing a child who responds, feels like you can grab the answer. It's bench to bedside, to bench,"*** said Dr. Gessler.

Noa's future development remains unclear, as the gene therapy is in early investigational stages. Still, *"It's really nice to be a part of the answers,"* said Lori. *"They see her, they learned from her, they saw what happened with her, and they're able to adjust it for the kids going forward. It's inspiring to be a part of the learning that's happening."*

"*You're working for Noa*," Lee Greenwood said as he thanked everyone. ***"It's not Baby 003; it's Noa."***

Read the full story of the Greenwood Family visit with UMCMS researchers here.

New findings from the first six CANaspire participants were presented at the 26th Annual Meeting of the American Society of Gene and Cell Therapy in May, as well as at NTSAD's Annual Family Conference. **Read more about findings from the study.**

NTSAD honored Dr. Gao in 2022 for his unparalleled achievements in gene therapy and Canavan disease at *Imagine & Believe.* Please read about this year's *Imagine & Believe* honoree further below in this newsletter.

Join NTSAD's Move for Hope Challenge

Each year, families and rare allies participate in NTSAD's Day of Hope to raise awareness and funds for research toward effective treatments for Tay-Sachs, Canavan, GM1, and Sandhoff diseases. Since 2011, families have been hosting their own Day of Hope events, raising $670,000 for research. **This year, we need your help in raising $55,000 for NTSAD's Research Initiative to fund cutting-edge research leading to potential treatments. There are many ways you can join the NTSAD Community in our 14th Annual Day of Hope.**

Join us on September 9th (or anytime) and Move a Mile for Hope. You can move, walk, dance, roll, stroll, or even drive for hope in your neighborhood. It's a great way to engage your family, friends, neighbors, and colleagues to participate in NTSAD's Day of Hope and our advocacy efforts.

If you're on Facebook, join the new NTSAD Move for Hope Facebook Challenge! The challenge will bring the NTSAD Community together as we Move for Hope and raise awareness in our communities. **Everyone who joins the Challenge and launches a Facebook fundraiser will receive NTSAD Miles for Hope purple shoelaces.** There also will be mini-competitions, incentives, and fun prizes for those who participate in the Challenge.

Join the Move for Hope Facebook Challenge

Once you receive your NTSAD shoelaces, lace up your shoes, and post a photo on social media of you and your rare family and friends to spread the word about Move a Mile for Hope on September 9th. Don't forget to tag NTSAD and use #NTSADDOH23 and #NTSADMoveforHope.

Have any questions? Reach out to our Development and Communications Manager, Sydnie Dimond at **sdimond@ntsad.org.**

We hope you will join us for Day of Hope, participate in Move a Mile for Hope, and the fight for treatments! **Since its inception in 2002, NTSAD's Research Initiative has awarded more than 69 grants and provided more than $4.1 million in funding.** The data generated in some of these projects funded by NTSAD grant opportunities were leveraged to obtain future funding from larger National Institutes of Health (NIH) grants, **resulting in more than $10 million toward finding effective treatments for Tay-Sachs, Canavan, GM1, and Sandhoff diseases and more than a dozen clinical trials in the last five years.**

Read more about NTSAD's recent research investments.

Give to Day of Hope and Support Research today!

You Can Host Your Own Day of Hope Event

Maybe you want to host your own Day of Hope. Over the years, families and rare allies have hosted bean bag/cornhole tournaments (The Gropp Family's tournament pictured above), sports competitions, bake sales, dance parties, walk/runs, rallies, and poker runs to raise awareness and funds for research.

Check out these special **Day of Hope events** hosted by rare families.

If you'd like to host your own personalized Day of Hope event in your community, please let us know, so we can help you with planning, including sending you materials, special Miles for Hope purple shoelaces, and giveaway items. Contact Development and Communications Manager, Sydnie Dimond at **sdimond@ntsad.org** for help with your event!

The Manning Family: 20 Years of Honoring Dylan

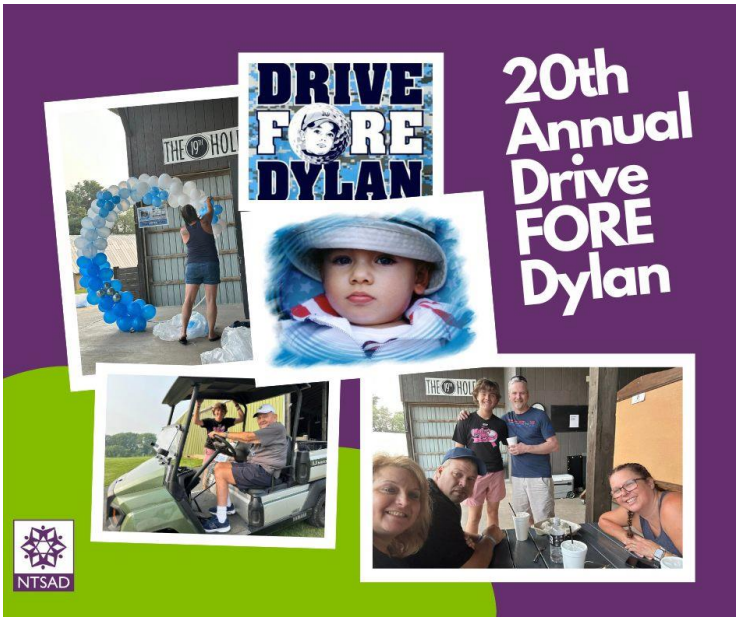
Dylan James Manning was born November 22, 2001, Sherri and Brian Manning's first child and the start of the family they would build together. Their hearts were broken when they found out that their beautiful boy Dylan was affected by infantile Tay-Sachs disease and would only be with them for a few, short years. **After being connected to NTSAD, Sherri and Brian wanted to find a way to help other families like theirs. In 2003, they planned their first Drive FORE Dylan Golf Tournament to raise funds for NTSAD.**

In 2006, Dylan passed away at the age of four, but his memory lives on through the Manning Family, which includes three other children, Brady, Devin, and Mia. Together, they continue to

spread awareness, kindness, and love in Dylan's name. In the past 20 years, the Mannings have touched so many people with their story and continued support of newly diagnosed families.

On June 30th, the Manning Family held the 20th Annual Drive FORE Dylan event. **With the help of their community, Drive FORE Dylan and the Manning Family have raised nearly $400,000 in support of NTSAD's family programs and research since 2003. It is an extraordinary part of Dylan's incredible and growing legacy.**

Learn more about the Manning Family and Drive FORE Dylan.



Stephanie Wright: Living with Late Onset Sandhoff Disease

In 2015, Stephanie started noticing weakness in her legs. It began around Easter when she was visiting her brother's family. Stephanie was jumping on the trampoline with her niece when she noticed it felt like her legs wouldn't support her.

"*When I first started experiencing symptoms, I just thought I needed to get to the gym*," says Stephanie. **"*I never in a thousand years thought I needed to go to the doctor.*"**

Stephanie was walking on a treadmill the first time it happened. She stepped off the treadmill and fell to the floor.

"*My legs just Jello-ed*," she says.

After three more embarrassing falls on the treadmill, Stephanie decided to leave the gym.

In June 2016, Stephanie went to see a neurologist in Houston for an electromyogram (EMG). The specialist had just read about Katie and Allie Buryk in *The New York Times* the week prior, and thought Stephanie's symptoms aligned perfectly with what he'd read.

The Buryk twins were diagnosed with Late Onset Tay-Sachs disease, which can be diagnosed with genetic testing. Stephanie underwent the testing and returned in September for one last test—one that could identify low levels of two vital enzymes—beta-hexosaminidase A (HexA) and beta-hexosaminidase B (HexB).

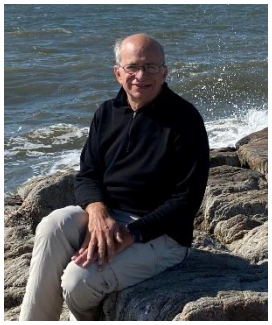
One month later, the neurologist called Stephanie. He had diagnosed her with Late Onset Sandhoff disease, which is very similar to Tay-Sachs disease.

Upon breaking the news to her family and friends, Stephanie fell into a depression that lasted about three months.

Then one day, she woke up and made a decision. **"*A geneticist told me I'd have a much larger chance at winning the multimillion-dollar lottery than having Late Onset Sandhoff disease. This must have happened for a reason. So, I've got to find my purpose,*"** Stephanie recalls.

Continue reading Stephanie's story and find out how she found purpose after her life-altering rare diagnosis.

Why I Give (to the Katie & Allie Buryk Fund)



Rick Boas is a brother, husband, father, and doctor. He is also a carrier of Tay-Sachs disease. Rick found out he was a carrier when he was an ophthalmology resident. As part of his training, samples of his blood and tears were tested. Once the tests were confirmed, he shared his carrier status with his siblings and his children.

Later, when he saw the *New York Times* article featuring Katie and Allie Buryk and their eight-year journey to receiving a diagnosis of Late Onset Tay-Sachs, he decided to support them and donate to the **Katie & Allie Buryk Fund for Research at NTSAD.**

"As a carrier and a doctor, I know it is important to humanize things and put faces to what is happening to real people like Katie and Allie. I strongly believe in doing what I can to support Katie and Allie in helping to spread awareness. Tay-Sachs and other rare genetic diseases are not on everyone's radar screen, even in high-risk groups with higher carrier rates. They are not discussed, and they should be. It is my pleasure to support Katie and Allie."

Thanks to many donors like Rick and his wife, Carol, The Buryk Fund has raised nearly a million dollars for research and funded several projects leading to clinical trials for Tay-Sachs and Sandhoff diseases. The Buryk Fund also gathers leading researchers and world renown thought leaders at an annual Late Onset Tay-Sachs and Sandhoff Think Tank to further innovate and advance research.

Please join Rick and Carol and donate to the **Katie & Allie Buryk Fund at NTSAD.** **Help the Buryk Family raise a million dollars for research!**

Give to the Katie & Allie Buryk Fund for Research Today!

Imagine & Believe 2023

Imagine & Believe with rare families and NTSAD at our signature event on November 9, 2023, from 6 to 9 pm at the Royal Sonesta Boston. *Imagine & Believe* brings together families, members of industry, clinicians, researchers, and rare allies for a reception, silent auction, and inspiring speaking program to raise critical funds for family services, programs, and research. Together, we envision a world with effective treatments for Tay-Sachs, Canavan, GM1, and Sandhoff diseases.

Each year we recognize an individual who makes a significant and positive impact on the rare disease community. **This year's honoree, Jayne Gershkowitz, Chief Patient Advocate at Amicus Therapeutics, is a trailblazer in patient advocacy and a remarkable individual who dedicated her career to championing children, adults, and families affected by rare diseases.**

Read more about Jayne Gershkowitz and her incredible advocacy.

Are you interested in sponsoring this event? **Check out the 2023 *Imagine & Believe* Sponsorship Opportunities.**

To sponsor *Imagine & Believe* or purchase an individual admission or digital program ad, please contact Sydnie Dimond, Development and Communications Manager at sdimond@ntsad.org.

Gottlieb Scholarship for Rare Siblings

Applications are now open for the Jeffrey Alan Gottlieb and Stanley N. Gottlieb Memorial Scholarship Funds, which provide financial support for college to healthy siblings of children and adults affected by Tay-Sachs, Canavan, GM1, Sandhoff or an allied disease.

In May 2005, Judy Gottlieb established two separate memorial college funds at National Tay-Sachs & Allied Diseases Association to honor her youngest son, Jeffrey Alan Gottlieb, who succumbed to Tay-Sachs in 1975, and her husband, Stanley N. Gottlieb, who passed away in 2001. **During the last 18 years, Judy has generously awarded scholarships to 56 siblings, some of whom have received multiple awards throughout their education.**

It is Judy's intent that the monies be distributed "for use in their college education, i.e., tuition, books, room and board."

Submit your application by **September 8, 2023,** for consideration this year.

Note: If you previously received a Gottlieb scholarship, and you are still in college, you may apply again!

Apply Today!

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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