NTSAD Community News

on Research, Community and Collaboration

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

November 2020

In this Issue

Giving Thanks to Our Community

As Thanksgiving approaches, we want to acknowledge our partners in this work - who have dedicated time, resources, and energy to finding treatments for children and adults affected by Tay-Sachs, Canavan, GM1 and Sandhoff diseases. We sincerely thank you.

NTSAD and our families cannot do this work alone. We continue pushing forward in honor of all who came before us and our entire community – each child, each adult drives what we do, and we honor them all.

After many ups and downs and tremendous perseverance by our community, there are three active clinical trials, and at least seven more drug development programs, a big change from just one year ago. Nearly 50 patients across the globe are enrolled in these trials, and there are an estimated 150 people involved in natural history or other clinical studies.

And recently, Sio Gene Therapies, formerly Axovant Gene Therapies, announced the FDA's approval to start the first-ever gene therapy clinical trial for infantile and juvenile Tay-Sachs and Sandhoff diseases!

Like many clinical trials, Sio Gene Therapies' achievement comes after many years of extremely hard work, in this case, by the Tay-Sachs Gene Therapy Consortium, a team of researchers from multiple institutions. During this time, NTSAD made more than $2 million in grants toward Consortium members' institutions working on gene therapy vectors, animal models, natural history studies, and more—all leading to the company's licensing of GM2 and GM1 gene therapy programs from UMass Medical School, which occurred less than two years ago.

Giving Tuesday

AmazonSmile for NTSAD

The CARES Act

There's still a long way to go before treatments are a reality, and the progress isn't as fast as our families need it to be. We also know that, while clinical trials offer a new level of hope--patients and families face complex decisions and logistics, and possibly another level of disappointment and grief. NTSAD remains poised to support and guide families as we navigate the new landscape of clinical trials, together.

Nonetheless, the progress is real, and we should celebrate it. These current studies and trials build upon many years of research and investments, not only by NTSAD, but by the NIH, medical centers, universities, and industry. I want to acknowledge the large teams from the industry sponsors and the clinical sites who support these studies.

Thank you for being a part of our community. May you be safe and well during the holiday season.

--Sue Kahn, NTSAD Executive Director

First GM2 Gene Therapy Clinical Trial to Begin Enrollment

Axovant Gene Therapies, recently renamed Sio Gene Therapies, prepares for first clinical trial for gene therapy for Tay-Sachs and Sandoff diseases after receiving Investigational New Drug (IND) clearance from the U.S. Food and Drug Administration (FDA) for AXO-AAV-GM2.

The study will enroll both infantile and juvenile patients with GM2 gangliosidosis. Enrollment will begin pending approval from Massachusetts General Hospital, Center for Rare Neurological Diseases and University of Massachusetts Medical Health Center's Institutional Review Board. The research study posting on ClinicalTrials.gov is pending review as well. The two-part trial will be led by Terence R. Flotte, M.D., Professor of Pediatrics and Dean at the University of Massachusetts Medical School, who will serve as principal investigator of the clinical trial.

Dating back to 2006, NTSAD has played an integral role by supporting this promising gene therapy research.

Read press release here.
More information for families here.

The company also received Rare Pediatric Disease Designation from the FDA for AXO-AAV-GM1, an AAV9-based gene therapy delivered via a single intravenous administration that is in Phase 1/2 development for GM1 gangliosidosis. AXO-AAV-GM1 is **the only gene therapy in clinical development for both infantile (Type I) and juvenile (Type II) GM1 gangliosidosis.** AXO-AAV-GM1 has Orphan Drug designation as well.

Learn more **here.**

Passage Bio's GM1 Gene Therapy Has Potential

The University of Pennsylvania's Gene Therapy Program supports the potential of Passage Bio's PBGM01 to **correct the underlying genetic defect of GM1 gangliosidosis.** Passage Bio announced publication of data in a murine model of GM1 gangliosidosis (GM1) demonstrating that a

single intracerebroventricular injection of an optimized adeno-associated virus (AAV) into the cerebral spinal fluid (CSF) resulted in significant expression of Beta-galactosidase (β-gal) in the brain and peripheral tissues, and **demonstrated dose-related reductions in neuronal lysosomal storage lesions, neurological impairment and improvement in survival.** These data support further development of PBGM01 as a potential therapy.

Read Passage Bio's press release **here.**

AllStripes and NTSAD Drive Data for GM1 and GM2 Research

AllStripes, formerly RDMD, is partnering with NTSAD and patients and their families to capture data via de-identified medical records that will be aggregated and leveraged to advance multiple research studies for both GM1 and GM2 to better understand how the disease affects patients, the natural progression of the disease, and the road to diagnosis.

In partnership with Cure GM1, data for GM1 patients has already been collected, and we will share the GM1 Research Insights report in a future newsletter.

Read more about the AllStripes GM2 project **here.**

Taysha Gene Therapies Partners with Invitae to Enable Earlier Diagnosis of GM2

Taysha Gene Therapies is partnering with Invitae to enable rapid access to genetic testing and earlier diagnosis of patients of rare diseases, including GM2 ganglisidosis (Tay-Sachs and Sandhoff diseases) via Invitae's Detect Lysosomal Storage Diseases (Detect LSDs) and Behind the Seizure ® programs. Read more **here.**

Becca Heringer Shares Her Story and Advocates for LOTS Community

Women's Health Magazine featured Becca Heringer who shares her story of accepting her diagnosis of Late Onset Tay-Sachs while embracing her artistic talent and entrepreneurship via her athletic outerwear company, **The Cosmic Project.**

"I am a person who has a lot of challenges when it comes to my speech and movement, but running an online business, none of that matters...I was nervous to share my story because I don't want people to look at me differently. However, it's more important to help raise awareness and be an advocate for this rare disease. It's crucial to me to speak for others who feel they haven't been heard, as they are impacted by something beyond their control...I realize that this disease is a part of me, but it doesn't define me." --Becca Heringer

Becca and her family are raising money for research for Tay-Sachs through the Heringer Fund.

Donate here.
Read the full article **here.**
Check out The Cosmic Project **here.**

Imagine & Believe Again

We remain grateful to **our sponsors and supporters of Imagine & Believe.** Together we raised more than $100,000 to sustain NTSAD's family programs and services.

We extend our gratitude to NTSAD families who during the event shared their stories and perspective, particularly Ryan Miller, Lorelei Sandoval, Sherri Sigel, and Justin Ungerleider.

We also sincerely thank Dr. Guangping Gao, gene therapy pioneer and researcher, and Dr. Florian Eichler, NTSAD Scientific Advisory Committee member and respected clinician, who along with Sherri, a NTSAD Mom, talked about the journey to a clinical trial for Canavan disease. **Watch the video here.**

Thank you to all the NTSAD families who shared their hopes for the future in a video produced by NTSAD Dad, Dan Redfield. **Watch here.**

We also want to share a video that highlights many incredible NTSAD families who support other families by providing understanding, compassion, and wisdom gained from their personal experience. **Watch here.**

If you missed the event, please watch **Imagine & Believe here.**

Donations are welcome.

Keep on Imagining & Believing! Purchase a holiday t-shirt and show that you still believe. A portion of proceeds go to NTSAD.

Purchase the Imagine & Believe T-shirt in red and green **here.**

Purchase the Imagine & Believe T-shirt in blue and white **here.**

10th Annual Day of Hope Raises $40,000 for Research

The 10th Annual Day of Hope once more highlighted the resiliency and unity of the NTSAD Community with families and companies raising awareness and funds while advocating for loved ones and rare disease patients.

Even during the ongoing pandemic, families and individuals across the country rallied to shine a light on the need for continued research toward effective treatments for Tay-Sachs, Canavan, GM1, and Sandhoff diseases. During our Season of Hope more than $40,000 has been raised thus far. We may be rare, but together, we are mighty.

Check out this poster from Jessie's Ride, a Day of Hope event, showcasing NTSAD kids!

Team Taysha shows their support for families during the Season of Hope!

#GIVING TUESDAY™

This year, Giving Tuesday, an effort to inspire philanthropy, is **December 1st**! We know due to the pandemic and economic fallout, not everyone can afford to give this year. We appreciate all the donors who continue supporting and sustaining NTSAD's important work.

Donate

amazon smile

This holiday season if you are shopping online with Amazon, consider choosing to support NTSAD as part of Amazon's charitable program, AmazonSmile.

It's super easy-- whenever you shop use this link to **AmazonSmile**, and Amazon will donate to NTSAD.

Have friends and family join you in supporting NTSAD through AmazonSmile by **posting to Facebook**

	and **Twitter**, too!

The CARES ACT: What You Need to Know

The CARES Act provides more incentive to give to charities by Dec. 31, 2020.

- There is now a Universal Charitable Deduction. Taxpayers who *do not* itemize their deductions can take a one-time deduction up to $300 for gifts made to a nonprofit like NTSAD.
- For 2020, there is no limit for individual deductions. The CARES Act suspends the 60 percent adjusted gross income limitations, so now cash contributions are fully deductible.
- Increases the cap on how much corporations may deduct for charitable gifts from 10 percent of taxable income to 25 percent.
- Waives for 2020 the required minimum distributions from retirement plans, such as pensions and 457 plans.

To learn more, go **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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