NTSAD Community News

Research, Community, and Collaboration

Supporting families is the center of everything we do…

September 2020

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Sanofi Genzyme Clinical Trial for Late Onset Tay-Sachs and Sandhoff

Sanofi Genzyme's clinical trial for Venglustat, a potential treatment for Late Onset Tay-Sachs and Sandhoff patients, has screened and officially enrolled its first two patients, and they have already received either the drug or the placebo per randomization requirements of the study. Sites at New York University Medical Center and Massachusetts General Hospital in Boston are actively recruiting patients and UCLA and National Institutes of Health will follow soon. Learn more about the clinical trial **here.**

Passage Bio Files IND for GM1 Clinical Trial

Passage Bio filed an Investigational New Drug (IND) for its infantile GM1 gangliosidosis gene therapy program this summer, a significant step in the process of drug development. Currently, the FDA is examining the device used in delivering the healthy gene and has put a temporary clinical hold on the trial, which is standard course for the FDA's oversight in ensuring every part of a clinical trial is safe. **Passage Bio expects to initiate the clinical trial in late 2020 to early 2021**. Read the full press release **here.**

Improved Delivery of AAV Gene Therapy in Tay-Sachs Sheep

In 2017, NTSAD awarded a grant to study the methods of delivering gene therapy to Jacob sheep, a rare breed of the animal with a naturally occurring form of Tay-Sachs. The project, which received a grant of $80,000, was led by principal investigator Heather Gray-Edwards, PhD, Assistant Professor of Radiology at UMass Medical School, and a member of the Horae Gene Therapy Center. While other gene therapy studies using intravenous (IV) administration in sheep with Tay-Sachs disease have been conducted, this was the first study to test the efficacy of cerebrospinal fluid (CSF) delivery in sheep. (CSF is the fluid surrounding the brain and spinal cord.)

The research showed that using CSF administration of AAV gene therapy is superior to IV administration finding all animals alive and well after more than two years of age at the conclusion of the study. Today, the sheep continue to live near-normal lives at the Farm at the Cummings School of Veterinary Medicine at Tufts University. The sheep are able to walk through the pastures and barn, rise to their feet unassisted, and eat normally. One ewe even had a lamb. Throughout the course of the grant, Dr. Gray-Edwards and her team established biomarkers in the animals reflecting those used in humans (ex. MRIs, CSF, etc.), helping to determine efficacy in future clinical trials. The success of the project has directly led to early-stage planning of a possible AAV clinical trial for Late Onset Tay-Sachs and Sandhoff disease in patients. To read more about Jacob sheep, please visit **this link.**

Wearable Technology Captures Symptoms of Late Onset GM2

NTSAD's Katie and Allie Buryk Research Fund supported a study to better capture disease symptoms of adults living with Late Onset Tay-Sachs and Sandhoff (GM2). **The six-month feasibility study evaluated patients' ongoing compliance in wearing digital health technology in order to capture continuous data remotely as well as changes often missed in between hospital visits.** The results of this natural history study, led by Cynthia Tifft, M.D., Ph.D., deputy clinical director for the National Human Genome Research Institute, found that patients had a high rate of compliance. The **recently published study** also set out to capture patients' perspectives on the impact of the disease and what is important to them via mobile Patient Reported Outcomes (mPROs). Since Late Onset forms of the disease progress slowly, the study did not capture any significant changes in symptoms; however, as the study was designed as a proof of concept, the potential of the wearable technology to capture long-term data will be expanded in future studies.

Sanofi Genzyme Fosters Collaboration Among U.S. Rare Disease Groups

This month, Sanofi Genzyme convened professionals from several orphan disease communities and advocacy groups at its 2020 U.S. Rare Patient Advocacy Leadership Summit. **NTSAD's Executive Director Sue Kahn presented on collaborating with industry for clinical research,** and she also gave advice about shifting in-person patient group conferences to virtual experiences along with co-presenter, Joslyn Crowe, Executive Director of the National Niemann-Pick Disease Foundation (NNDPF).

Aspa Therapeutics Developing Gene Therapy for Canavan

Aspa Therapeutics will host an educational webinar for families affected by Canavan disease. Professor Guangping Gao, a pioneer in gene therapy who developed Aspa Therapeutics' investigational gene therapy program, along with an expert panel, will discuss Aspa's clinical development program. There also will be a general discussion about clinical trials and an opportunity to ask questions.

Families must **register** for the webinar to receive the link to participate on **Wednesday, September 16, 8 pm Eastern time/5 pm Pacific.** Questions? Email **patientadvocacy@aspatx.com**

Social Security Administration Recognizes GM1 for Disability Insurance

NTSAD's Director of Family Services Diana Pangonis successfully advocated for families with infants and juveniles diagnosed with **GM1 Gangliosidosis to be included among the list of conditions to receive Compassionate Allowances from the Social Security Administration.** The program

essentially fast-tracks applications for Social Security Disability insurance due to rare diseases that affect children. GM1 is officially included along with Canavan, Sandhoff, and infantile Tay-Sachs. NTSAD's request to add juvenile Tay-Sachs is pending. You can learn more about the Compassionate Allowances program **here**, and read the full press release **here**.

Foundations Rise to Support Families During COVID-19

The Child Neurology Foundation (CNF) awarded NTSAD with a Rising Tide grant to sustain NTSAD's ability to respond to rare families' needs during COVID-19. The $10,000 in financial support as well as the **resources** CNF provides help families when they need it most.

The Hamilton Company Charitable Foundation renewed its support of NTSAD's Family Services to ensure families receive critical individualized support as well as access to the entire NTSAD community via peer support groups. Connection and community are vital to families with newly diagnosed members and for families who are caretaking as well as grieving. All families struggle with feelings of isolation, which are exacerbated by the ongoing pandemic.

The Power of Connection Among NTSAD Families

The NTSAD family network worked its magic once again. This time in San Diego. Maria Vasquez, who contacted NTSAD after learning her 18 month-old son, Nathan, has infantile Sandhoff disease. Isolated and scared, Maria had no information about Sandhoff or what the diagnosis meant for her son. Diana Pangonis, NTSAD's Director of Family Services, provided Maria with resources and connected her with other NTSAD families in the area. Soon Lorelei Sandoval and her son Isaac, who was diagnosed several years ago with Tay-Sachs, visited with Maria and Nathan at the local children's hospital offering in-person (and socially distanced) support. Lorelei also gifted Isaac's previous adaptive stroller to Nathan! Lorelei, along with other families, reached out and offered Maria support, easing the isolation and fear that follow a diagnosis of a rare disease.

In addition, NTSAD provided Maria's family with a grant from Emma's Fund for Families, established with NTSAD to enable families' ability to experience memory-making opportunities in honor of the fund's namesake, Emma Artinian. The Vasquez family used the grant toward an annual membership to the San Diego Zoo. Now, Maria, Nathan and her other son Christopher can visit the world renown zoo as often as they like!

When Diana reached out to Maria about this story, Diana was surprised by Maria's heartfelt and candid response. ***"I think your support has been more helpful than my family members, because they really do not understand or they don't want to. They haven't been through this, and [the other NTSAD families] Jan Marquez, and Lorelei, and you have been really helpful to me in so many ways just by talking to me, hearing me out, and giving me advice...*** *"It's still really hard [for me] to share pictures of Nathan. [But] here's a quote [about faith] that I think is perfect for what we are going through, and all the help that NTSAD has given me...maybe you could share it [to show how] I feel for this program and finding you, Jan, Lorelei, and this amazing, beautiful program. And that 'I'm not the only one.'"*

To learn more and **donate** go to Emma's Fund for Families.

Day of Hope: Join Team M&M!

Each year Mandy and Jeff Ronaldson host a NTSAD Day of Hope Event, named Team M&M in honor of their daughters Mollie and Madelyn. **The Miracles for Mollie and Madelyn campaign is an effort to raise awareness and funds for research to find treatments for their daughters who have Juvenile Sandhoff disease.** Recently, Mandy appeared on the radio and talked about her girls and her family, and her community's efforts in hosting a virtual walk/run. **Listen to interview here,** watch a video of the girls, and consider participating in Team M&M's virtual 5k walk/run on September 12. Every participant gets a Team M&M t-shirt, and can order one, even if you don't walk or run. Proceeds from t-shirts support their efforts and advance research toward treatments. Order yours **here** before they run out.

Show your Support: Wear that You Care for Rare!

If you'd like to join NTSAD's community of supporters for the 10th Annual Day of Hope by showing that you #careforrare you can visit the NTSAD Bonfire store and purchase your own Day of Hope 10th Anniversary T-SHIRT and NTSAD FACE MASK.

Get your 10th Anniversary NTSAD Day of Hope t-shirt **here**!

Stay safe and order your NTSAD 10th Anniversary Day of Hope face mask **here**!

NTSAD Team Updates: Thank You Ingrid and Welcome Sydnie

The NTSAD Community recognizes Ingrid Miller for her years of service and commitment to NTSAD as well as for returning to NTSAD last November to resume her role as Development and Administrative Associate to aid NTSAD in stewarding donors' gifts and supporting NTSAD families. Having wrapped up a second stint with NTSAD, Ingrid will resume her volunteer work with disabled adults and spend more time with her family. Sue Kahn, Executive Director shared, ***"NTSAD will always be indebted to Ingrid, and we thank Ingrid for supporting and helping with onboarding the new Development and Communications team. Ingrid will always be a part of the NTSAD family!"***

Last month, NTSAD welcomed Sydnie Dimond as the new Development and Communications Associate. Sydnie, a graduate from the University of Massachusetts, Amherst with a degree in nonprofit management and development, has a calling for supporting families affected by critical illness and rare diseases. Previously, Sydnie worked with the March of Dimes and the Make-A-Wish Foundation, as well as with PR firm The Castle Group. ***"I am deeply humbled by NTSAD's families' experiences and strength. I hope I can be of help to this amazing community and serve NTSAD's powerful mission."***

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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