NTSAD Community News

Research, Collaboration, and Community

Supporting families is the center of everything we do…

May

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In this Issue

Dear NTSAD Community,

NTSAD's powerful mission is to lead 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. Recently we updated our vision statement, which truly reflects our shared hopes for the future as well as our ongoing journey. I am excited to share it with you.

NTSAD envisions a world in which Tay-Sachs, Canavan, GM1, and Sandhoff diseases are no longer fatal or debilitating. At NTSAD, we will:

–Above all else, provide compassionate support, advocate for patients and their families, and promote early diagnosis and prevention.

–Empower, educate, and connect affected families and individuals.

–Serve as the preeminent resource for families, industry members, researchers, and clinicians.

–Direct, promote, and invest in research to accelerate the development of treatments and cures.

–Act as a leader within the rare disease community.

In the 65 years since NTSAD's founding, one of our greatest accomplishments was launching the Research Initiative in 2002. Since its inception NTSAD's Research Initiative has awarded more than $4 million in grants that have been leveraged to more than $30 million in additional funding. Under the direction of our new Research Director, Valerie Greger, PhD, the Research Initiative, which had been paused since 2017, was relaunched earlier this year. This summer, we will award more funding to new meritorious research.

Over the years, the NTSAD Community has experienced many bumps in the road to clinical trials. Recently, the biotech sector has suffered some financial woes, but as the data remains positive, we remain cautiously optimistic about the future of these trials, while recognizing more challenges lie ahead as we develop effective treatments for Tay-Sachs, Canavan, GM1, and Sandhoff Diseases.

In fact, while attending the American Society of Gene and Cell Therapy (ASGCT) 25th Annual Meeting in Washington D.C. this week, Diana Pangonis, NTSAD's Director of Family Services, and I had the pleasure of meeting in-person with principal investigators, researchers, and industry partners who have made our Community stronger and remain committed to driving research and clinical trials. At the conference, we heard updates on relevant research and trials, and made plans for future collaborations. We are grateful to the American Society of Gene and Cell Therapy for bringing patient groups, researchers, and doctors together for three days of education and collaboration. I know that great progress is on the horizon.

Sincerely,

Kathleen M. Flynn
Chief Executive Officer

P.S. Read more about the ASGCT Annual Meeting further below.

Passage Bio Reports Positive Data for GM1 Clinical Trial

At the American Society of Gene and Cell Therapy (ASGCT) Annual Meeting Passage Bio presented positive data on the company's GM1 clinical trial including development improvement for both patients by study investigators and caregivers. In addition, the low dose of the gene therapy, PBGM01, was well tolerated, and the company continues to enroll patients in Global Phase 1/2 study.

Read Passage Bio's press release.

GM2 Clinical Trial Update

In late April, Sio Gene Therapies announced the company's termination of its licensing agreement for gene therapies with the University of Massachusetts Chan Medical School (UMCMS). Immediately following the news, NTSAD reached out to the research teams at UMCMS and the National Institutes of Health (NIH), who assured us they remain committed to research involved in the trial. After

receiving that communication, NTSAD convened a meeting with other patient advocacy leaders and drafted a joint statement that was shared among the respective patient communities.

Last week, NTSAD's leadership met with researchers and administrators at UMCMS, who reported they are working with Sio to ensure a smooth transition so that the GM2 trial continues. UMCMS is negotiating with Sio to secure patient data and samples, the transfer of the IND (Investigational New Drug) with the FDA , and obtaining the remaining vector (gene therapy "drug"). It is our shared hope that another company will make the investment to take over the trials.

Read the joint statement from NTSAD and patient groups.

44th Annual Family Conference July 7-10, 2022

Thinking about attending NTSAD's Annual Family Conference? Individuals and families coping with a diagnosis of Tay-Sachs, Canavan, GM1 and Sandhoff diseases are encouraged to register and attend sessions on caregiving, family planning, living with grief, and updates on the latest research and clinical trials across all NTSAD's rare diseases. Powerful connections are made at peer group meetings, meals, in the family lounge, and during activities for affected and healthy children. See what it's all about and watch a video from the 2019 Conference.

Consider joining NTSAD at the 44th Annual Family Conference from July 7 to 10 in Denver, Colorado.

Register Now.

NTSAD's offers financial assistance for families in need! **Apply for a Helping Hand Grant here.**

It's not too late to support the conference.

Thank you to all our 2022 Conference Sponsors!

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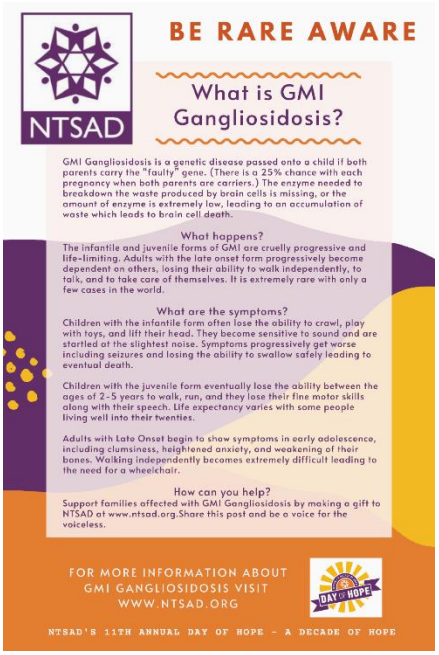
*NTSAD Board Member

International GM1 Gangliosidosis Awareness Day: May 23rd

International GM1 Gangliosidosis Awareness Day is Monday, May 23rd. Join NTSAD and our partners as we spread awareness of GM1 gangliosidosis. Participation is easy.

- If you care for an affected loved one, share your story and a picture on social media.
- Check out Cure GM1's **social media kit** for ideas. This kit includes the GM1 day logo in multiple languages.
- **Add the GM1 Awareness Day frame to your Facebook profile picture.**
- Share NTSAD's GM1 Be Rare Aware Infographic on social media. (Right click to save the graphic to the left.)
- Follow NTSAD on **Facebook**, **LinkedIn**, **Twitter**, and **Instagram**, and encourage others to do so, too. Share NTSAD's awareness posts on May 23rd.
- **Donate to support affected families.**
- Create a **Facebook** or **Instagram** fundraiser to encourage your friends to donate (link to instructions).



Maryland and Iowa Pass Newborn Screening Legislation

In the last month, both Maryland and Iowa have passed lifesaving newborn screening bills. The laws, referred to as Recommended Uniform Screening Panel (RUSP) alignment legislation, will implement a two-and-a-half-year timeline in which screening in the state must begin for new conditions added to the federal RUSP. Iowa and Maryland are the second and third states to adopt RUSP alignment legislation this year, with ten states overall enacting this newborn screening legislation. These bills will help enable earlier treatment and better outcomes for hundreds of babies. Thank you to the EveryLife Foundation for their tireless advocacy that made these bills a reality!

Read about Maryland's Newborn Screening Law.

Read about Iowa's Newborn Screening Law.

Recognizing Courageous Mothers

Every May, we recognize and celebrate moms. NTSAD acknowledges that Rare moms have a more complicated relationship with Mother's Day. Years ago, Bereaved Mother's Day was created and is held on the first Sunday in May to acknowledge mothers who have lost a child and people who have lost mothers. Every day NTSAD acknowledges the courage of Rare mothers, many who face the challenges of caring for an affected child or adult, wrestling with anticipatory grief and worry, and learning to carry the weight of loss in their hearts. A mother's love is forever. Join NTSAD as we recognize all Rare moms for the strength, perseverance, and love they demonstrate every day.

NTSAD Community and Board Members Advocate for Patients and Families

Both NTSAD's Board President Staci Kallish, DO, and Florian Eichler, MD, Director of the Center for Rare Neurological Diseases at Massachusetts General Hospital (MGH), participated in the MGH's World Medical Innovation Forum in Boston earlier this month. In a Q &A session with Tazeen Ahmad, Managing Director, Global Research, Bank of America Securities, Dr. Eichler was asked what criteria should be used to determine the safety of treatments and the tolerability of side effects, particularly when it comes to diseases that are often fatal when untreated.

"Safety is always first and foremost on our mind as treating physicians who are engaging with patients"* Dr. Eichler said. *"As we work in this space, we're constantly counseling families to weigh the risk-benefit of treatment to understand what to expect."

An NTSAD Dad expressed his heartfelt appreciation during the panel for Dr. Eichler's efforts to identify and test potential treatments and his openness about the risks involved.

"My family, we certainly know the pros and cons. We're going into this with our eyes wide open. But we know what the path is when there is no hope, there is no treatment, and that path is awful. To have something, anything, where we can give our child some hope—it was an easy choice, honestly." *-NTSAD Dad*

During the Professional Patient Advocates in Life Sciences (PPALS) training program another NTSAD Board Member Jamie Ring reminded participants what Henri Termeer, Founder of Genzyme, once said, *"If you do what is right for patients, the business will follow."*

ASGCT 25th Annual Meeting

NTSAD's CEO Kathy Flynn and Director of Family Services Diana Pangonis attended the American Society of Gene and Cell Therapy (ASGT) meeting in Washington, DC the week of May 16th where they heard updates on relevant research and trials and made plans for future collaborations.

The week kicked off with an incredible workshop, "What to Expect as a Participant in a Clinical Trial." Speakers touched on topics ranging from designing clinical trial, clarifying what informed consent means for families, and weighing the risks and benefits of participation in a clinical trial. These topics will be explored further with NTSAD families in an upcoming issue of *Lifeline* newsletter and future webinars.

Several presentations focused on the future of gene and cell therapies for Tay-Sachs, Canavan, GM1, and Sandhoff diseases. All were hopeful and encouraging.

Throughout the week, Kathy and Diana met with key stakeholders in the NTSAD Community, including Florian Eichler, MD, Mass General Hospital; Cynthia Tifft MD, PhD from the NIH; Kevin Romer, NTSAD parent and founder of the Mathew Forbes Romer Foundation; Jagdeep Walia MD, from Queens University; Marie Trad, MD, Chief Medical Officer at Lysogene; and Heather Lau MD from UltraGenyx. In addition, new connections were made that enhance the NTSAD Community and advance research.

Thanks again to ASGT for bringing patient groups, researchers, and clinicians together for three days of education and collaboration as well as inviting NTSAD to attend compliments of ASGCT.

Get Free at-home COVID-19 Tests

Every home in the U.S. is eligible to order a third round of free at-home COVID tests. It's quick and easy to sign up. Have tests sent to your home so you can have the tools to know your safe to travel and attend the NTSAD Annual Family Conference. Be prepared and order yours today!

Order free COVID tests here.

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