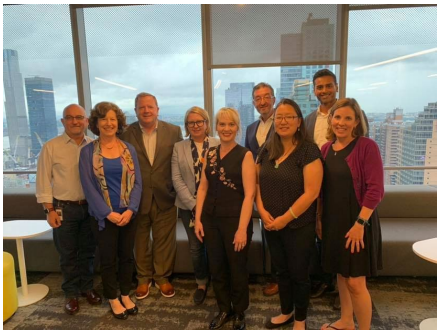
September 2019

Clinical Trial Updates

GM1 Gene Therapy

It was big news when it was shared that the first patient was treated with the first ever gene therapy for GM1 Gangliosidosis. It warranted a tea party with the team at NIH led by NTSAD's longtime Scientific Advisory Committee member (and former chair), Cynthia Tifft, MD, PhD.

We look forward to future updates as the trial progresses. Read the article from NIH Clinical Center News **here**.



GM2 Gene Therapy

Axovant Gene Therapies has kept NTSAD and Cure Tay-sachs Foundation (CTSF) informed as they continue to work toward filing the Investigational New Drug application (IND) - an important and necessary step toward opening a GM2 clinical trial. They invited NTSAD, CTSF and Cure GM1 to a town hall meeting in July to speak with the Axovant team to keep them connected to the family community. We hope to have more updates in the coming months.

Canavan Gene Therapy

On Wednesday, the 28th of August, Canavan Foundation, Canavan Research Illinois and NTSAD met with Aspa Therapeutics to discuss updates on their gene therapy program. Aspa reported they have been in discussions with FDA and that they remain on track for a gene therapy treatment trial to be open for enrollment in 2020. Agreement has been reached on the preclinical requirements, and manufacturing is on schedule.

Read the update in its entirety **here**.

Retrotope

At the May 2019 American Academy of Neurology Scientific Sessions, Dr. Robert Molinari presented Retrotope's poster presentation entitled: "RT001 to Treat Neurodegeneration--Case Study: Improvement in a Single Patient with Late Onset Tay-Sachs Disease (LOTS).RT001 is a di-deuterated isotope of linoleic acid (D-LA) that is readily incorporated into membranes and prevents oxidative damage. Read the full poster abstract **here**.

Site at Mayo Clinic Opened for IntraBio IB1001 Clinical Trial

A second site at the Mayo Clinic in Rochester, MN, has opened for the IntraBio clinical trial for N-Acetyl-L-Leucine for GM2 Gangliosdisosis (Tay-Sachs and Sandhoff Disease). To learn more about this clinical trial, visit **here**.

*NTSAD's first webinar featured a presentation by IntraBio and a researcher who worked on the compound, N-Acetyl-L-Leucine. View the full webinar **here**.*

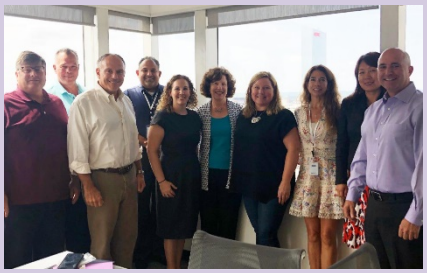
Breaking News: IntraBio announced their clinical trial for *N-Acetyl-L-Leucine for GM2 Gangliosdisosis* has opened for recruitment in the United Kingdom. Sites will be at the Royal Manchester Children's Hospital and the Salford Royal NHS Foundation Trust. Read the full press release **here**.

Natural History Studies to Inform Clinical Trials

Passage Bio announced their launch of a **GM1 Gangliosidosis** natural history study for the infantile and juvenile forms of the disease. Read the full press release **here**.

Aspa Therapeutics launched *CAN-Inform* to begin the process of building a comprehensive natural history study for **Canavan disease**.

To enroll in the natural history study, *CAN-Inform*, a family will need their child's medical record. Aspa is providing a free service to families to collect the medical record. 15 families have signed up for record retrieval so far. For those families who are able to travel to the clinical site, Aspa is providing travel support. It is important for families to sign up soon as it can take some time to obtain all records. Families who have lost a loved one are also encouraged to participate. To sign up for this service, please call 1-833-764-2267 or **CanavanMedRec@veristat.com**.



NTSAD Meets the Team at Passage Bio

Senior management at Passage Bio invited Sue Kahn, Executive Director, and Staci Kallish, DO, NTSAD's Board Chair, to Philadelphia to meet with the team to learn more about NTSAD, our community and how we can work together moving forward. Meeting and consulting with industry partners has become a bigger part of what we do as we advance clinical trials.

JScreen and Next Generation Sequencing (NGS)

NTSAD has partnered with JScreen for several years and co-funded the work leading to this newly published study, that has been highlighted in the press as we recognize September as Tay-Sachs Awareness Month.

“NTSAD was a pioneer in the prevention of Tay-Sachs in the Ashkenazi Jewish community through carrier screening programs and education. 'While the utilization of Tay-Sachs carrier screening has been impactful and regarded by many as a model public health genetic screening initiative, affected babies are still being born to Jewish, interfaith and non-Jewish couples who are not offered testing,' said Staci Kallish, DO, NTSAD Board President."

For more information on the study, or to review the entire article, please visit **here**.

Co-funded with our affiliates the Cameron & Hayden Lord Foundation and Mathew Forbes Romer Foundation, and the New York Area and Evan Lee Ungerleider Funds of NTSAD.

Grant Updates from NTSAD-funded Research Projects

When NTSAD funds any research project, it is a requirement to receive timely progress reports throughout the two-year grant. We are pleased to share updates from the following researchers on their funded projects. You can read the lay summaries here.

Alessandra d'Azzo, PhD
Department of Genetics
St. Jude Children's Research Hospital

Role of Plasma membrane-ER Contact Sites in GM1-mediated Neuronal Cell Death

These studies may also reveal novel or alternative therapeutic avenues for patients affected by GM1 gangliosidosis.

READ MORE

Heather Gray-Edwards, PhD, DVM
Department of Radiology
UMass Medical School

READ MORE

Minimally invasive delivery of AAV gene therapy in the Tay-Sachs sheep

The goal is to determine the efficacy of AAV gene therapy using established biomarkers after CSF delivery.

Xuntian Jiang, PhD
Metabolomics Facility
Washington Univ in St. Louis

READ MORE

Oligosaccharide Biomarkers for Disease Progression and AAV Therapeutic Efficacy in GM1 Gangliosidosis

The goal is to determine the efficacy of using H3N2b as a biomarker to determine treatment efficacy which will provide a much-needed tool for assessing GM1 disease severity and therapeutic efficacy.

Ninth Annual Day of Hope

We want to **thank our families**, their friends and communities for rallying and raising funds for research since 2011 as we all look toward that Day of Hope! **Together, they've raised over $440,000 in eight years!**

Good luck to all who are having events this weekend and the following weeks!

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