



NTSAD News



Aspa and Axovant Press Releases

Aspa Therapeutics shared data indicating the most suitable method of delivery for their Canavan gene therapy program at the European Society of Gene and Cell Therapy (ESGCT) on October 22, 2019. Read the full report [here](#) and the full abstract poster [here](#).

At the same meeting, **Axovant Gene Therapies** issued a press release ([here](#)) about their presentation of initial data gathered from two patients who were treated as part of an expanded access study for GM2 gene therapy.

We will continue to keep you updated on any news that we receive from Aspa and Axovant via our e-news, Facebook and Twitter.



Early Reports: GM2 Gene Therapy

WBUR and the CommonHealth blog ran a post highlighting the early but exciting data from two patients treated at UMass Medical Center (separate from Axovant). The news is cautious but hopeful. NTSAD is proud to have been a part of this landmark gene therapy program since its early beginnings 11 years ago.

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[CommonHealth](#)

Read the full blog post [here](#).

Canavan Natural History Video Launch

Canavan Foundation, Canavan Research Illinois and NTSAD came together to collaborate on an animation explaining a natural history study and the launch of a comprehensive natural history study led by Aspa Therapeutics for Canavan disease.

For more information about the study, visit the CANinform website [here](#).



Global Genes Rare Patient Advocacy Summit

Sue Kahn, Executive Director, and Diana Pangonis, Director of Family Services, attended the RARE Patient Advocacy Summit in September where they divided and conquered to gain insights, meet with rare advocates, and learn new perspectives. Over 1,000 patient advocates from around the world were in attendance.

With our rare community in the midst of one gene therapy trial and on the cusp of several more, NTSAD is positioning itself to support families in many ways. Highlights included:

- * Meeting an Indian delegation led by Prasanna Shirol at ORDI and meeting a parent, Deepa Kodali whose son has Juvenile Tay-Sachs. We hope to collaborate with ORDI to support a growing number of families in India.
- * Learning about a new initiative, Our Odyssey, created by Seth Rotberg, with the goal to provide a safe space for young adults affected by rare diseases to connect via in-person meetups and virtually.
- * Participating in the first in-person Rare Foundation Alliance meeting to learn about a soon-to-be-launched resource that will be invaluable as we navigate clinical trials.
- * Attending the Cure GM1 Foundation's GM1 Family Meeting on Friday, September 20th was a wonderful opportunity to hear updates on the latest in GM1 research with updates on the clinical trial programs in process and in development.

One of the speakers from another rare disease group shared her analogy to getting to treatments - it is a marathon. You have to be focused at the start line, keep going as the run isn't easy, but keep alive drive to get to the finish line.

Late Onset Think Tank | October 3-4, 2019

The Second Annual Late Onset Think Tank meeting was recently hosted by Alexis Buryk, parent of Katie and Allie who are both affected with Late Onset Tay-Sachs.

Late-onset Tay-Sachs and Sandhoff diseases (LOTSS), caused by b-hexosaminidase A deficiency due to mutations in HEXA or HEXB genes respectively, are slowly progressive yet relentless diseases with no effective therapy. Disease progression is variable, even within sibships, and has not been well characterized. Large animal models have been helpful in characterizing disease pathogenesis, but more work is needed. Likewise, biomarkers that correlate with disease progression have been lacking.

In response to the need for filling in these research gaps in LOTSS and prompted by Alexis Buryk and the Katie & Allie Buryk Research Fund of NTSAD, co-chairs Dr. Cynthia Tiff and Dr. Steven Walkley organized the LOTSS Think Tank. The LOTSS Think Tank brings together expert clinicians, translational and basic scientists to strategize a rapid path forward to address the unanswered questions.



Florian Eichler, MD, Dan Ory,
Xuntian Jiang (Left to right)



Thirteen experts attended the second annual LOTSS meeting in Hilton Head, SC, just a few weeks ago, one year after the initial LOTSS Think Tank meeting. The participants identified key needs and available resources for moving forward in each of these areas. A shared site for notes, papers, slides and documents was established as well as the beginnings of a virtual biorepository. Subsequent smaller meetings were held at the WORLD Lysosomal Diseases meeting and at the Gordon Conference of Lysosomal Diseases. Note taking, coordination of calls and management of the DropBox site became the responsibility of a post baccalaureate student at the NIH who became the coordinator for the LOTSS Think Tank. The costs of the LOTSS Think Tank is underwritten by the Katie & Allie Buryk Research Fund of NTSAD.

All of NTSAD diseases could benefit from better natural history, animal models and biomarkers of disease progression. We believe that this Think Tank model can expand within LOTSS and may also serve as a template in our other diseases.

FoCUS: An MIT Initiative

While gene therapy clinical trials and ultimately approved treatments are highly anticipated, we know that these treatments could often involve high up-front costs.

An initiative running at **MIT NEWDIGS** named **FoCUS (Financing and Reimbursement of Cures in the US)** is examining new innovative financing and reimbursement models for these types of treatments with the goals of (1) patient access for needed treatments; (2) financing and payment models for public and private payers; and (3) the sustainability of innovation by biotech / pharma. NTSAD was invited to participate in patient research as part of this project to better understand patient perspectives of medical financial burdens.



NTSAD reached out in August to families and affected individuals to invite them to participate. Responses were deidentified and shared and are being used by the FoCUS research team. This is part of a broader research effort that will encompass more patient organizations and will result in a publication in the spring.

NTSAD Events



Imagine & Believe: An Evening to Benefit NTSAD is fast approaching with a stellar evening planned for October 30th. We're thrilled to be able to honor our longtime friend, Dr. Cynthia Tiff, and recognize the incredible work she has pioneered over the years.



The **Ninth Annual Day of Hope** was a resounding success with over \$58,000 raised by families coast-to-coast! Their efforts have now raised over \$500,000 for research and family support since 2011.

Our thanks... to the Kliger family and their friends for hosting their **11th Annual 'Fore' Jack Memorial Golf Outing** on October 7th in New York.

We also want to thank **Ed Goldstein and Melanie Flamenbaum** for organizing their event featuring astronaut John Grunsfeld on October 13th in Denver, Colorado.



Their efforts and commitment to honor their children by raising awareness and funds for the work NTSAD does every day is appreciated beyond measure.



A gift to NTSAD is a gift to families and always appreciated.

Give Today.



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