



National Tay-Sachs & Allied
Diseases Association, Inc.

Friday, SEPTEMBER 16, 2016
NTSAD Monthly Research Review

DAY OF HOPE: SEPTEMBER 17, 2016

NTSAD designates one day in the year (although hope can be recognized anytime) for the community to rally and raise funds for research. Events are held from coast to coast ranging from motorcycle rides, yard sales to beanbag tournaments. Since 2011, over \$168,000 has been raised by families and their friends and communities.

This year we hope to surpass \$200,000!

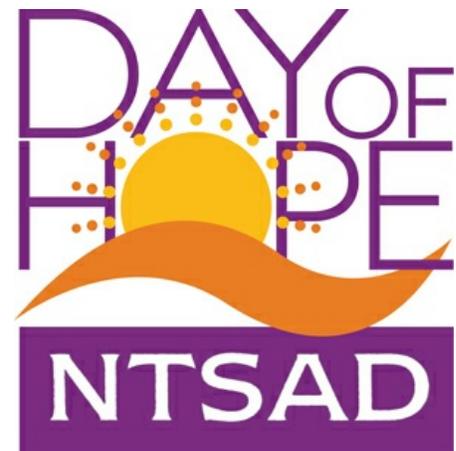
Several researchers shared what NTSAD's support means to them as they work toward finding treatments for Tay-Sachs, GM1, Sandhoff, Canavan and related genetic disorders.

David Radin, BioStrategies

"The grant support we received from NTSAD has enabled our research on therapeutic drug development for GM1 to move forward in a new innovative direction by providing seed funding for us to pursue a high risk high reward research idea that would not have been possible without this assistance from NTSAD.



This grant has more broadly inspired us by providing an opportunity for us to join the NTSAD family, attending NTSAD annual meetings to present our research to patients and families, and sharing our hope in the potential of our research to bring significant new therapeutic options for these patients. The grant funding from NTSAD has also strengthened our ability to raise follow up grant funding from NIH and to significantly accelerate the pace of our GM1 research. We are extremely grateful to NTSAD families for all of these



Make a Gift [HERE](#)
for Research

The field of rare disease research is expanding. New technologies are developing. Really smart people are dedicated to hurdling the neurological roadblock so we can have treatments for these diseases. **All of these efforts need funds to keep moving forward to the finish line!**

All events held for **Day of Hope** go to NTSAD's **Research Initiative** Fund which allows



benefits."

Click [here](#) to read the latest update on this research.

Doug Martin, Auburn University

"I'm extremely grateful for NTSAD's support of my research program on GM2 gangliosidosis, for many reasons. They boosted my confidence by funding early gene therapy studies, which led to federal and corporate support of several million dollars. They helped maintain and study our GM2 animal models - in fact, the sheep model of Tay-Sachs disease might not still exist without NTSAD's support. Most importantly, the research that they funded has moved us much closer to a real, effective treatment for Tay-Sachs and Sandhoff disease. Thanks NTSAD!"



As Doug mentioned, the most recent NTSAD Research Initiative grant to Doug's work, and the initial results generated, led to a five year multi-million dollar NIH grant to optimize vector delivery to the brain, spinal cord and peripheral organs to treat CNS and peripheral disease simultaneously.

Eric Sjoberg, OrPhi Therapeutics

"The \$25,000 grant that OrPhi Therapeutics, Inc. received from NTSAD's Katie & Allie Buryk Research Fund for developing an animal model for Sandhoff Disease with a disease causing missense mutation, has allowed us to secure \$150,000 in SBIR funding for initial dosing studies for pharmacological chaperone therapy of GM2 gangliosidoses. Our pharmacological chaperones are small molecules that bind and stabilize normally unstable mutant Hex (and likely HexA) increasing the activity of these deficient enzymes to a sufficient level to degrade toxic products that build up in these diseases. Pharmacological chaperones enter the CNS, making them useful for treating the CNS manifestations of the disease. Based on this animal model we believe we will be able to obtain a phase 2 SBIR worth up to \$1MM to demonstrate in vivo efficacy data leading to an IND filing. So for an initial \$25,000 investment, these funds may allow for a 30-40 fold increase in research funding



NTSAD to make grants to those really smart people.

To see if an event is happening near you, click [here](#) for HOPE.

So...let's make this a big year. Contact Joan at NTSAD to make things happen at joan@ntsad.org.

LATE ONSET STUDY A Pioneer Group of Eight



Late-onset Tay-Sachs and Sandhoff disease patients receive their Aparito wearable devices and phone apps as part of a 6 month pilot study to monitor activity and record patient-related outcome survey responses on activities of daily living and general well-being. With information obtained from these 8 pioneers Dr. Tift and her study team hope to identify important outcome parameters that can be used for clinical trials. The study is being funded by NTSAD through a generous donation from the Katie & Allie Buryk Research Fund of NTSAD.

**MILLION DOLLAR BIKE RIDE
Request for Proposals**

LAST DAY TO SUBMIT

The Request for Applications (RFA) on the Orphan Disease Center website, [here](#).

(relative to the initial grant) that could ultimately lead to a therapy to treat GM2 gangliosidosis."

Click [here](#) to read the summary of Eric Sjöberg's work.

THE GENEROSITY OF FRIENDS

An excerpt from Alexis Buryk's *Huffington Post* blog

If you want to go fast, go alone, if you want to go far, go together. - African proverb

"Due to an incredibly generous friend, Peggy Furth, the premier medical institution in the country, The Mayo Clinic, has launched the Mayo Clinic Lysosomal Disease Initiative. This is the first of its kind in the country. Peggy's generosity, persistence and can-do enthusiasm made this happen. Her gift to The Mayo Clinic came with a proviso; the money had to be used to find a cure for Tay Sachs.

The Mayo Clinic has assembled an outstanding team to focus on finding a cure. Tay Sachs is just one disease that falls under the lysosomal disease category. This will not only help Tay Sachs patients but also those afflicted with Pompe, Sandhoff, Gaucher and Niemann-Pick to name a few. We are beyond excited and filled with hope for all the possibilities for my children as well as others with these rare genetic disorders.

As for my other family and friends, we would not have been able to raise over \$270,000 for the Katie & Allie Buryk Research Fund to find a cure for Late Onset Tay Sachs. We have funded 4 research grants with some of the money and thanks to all of them, we will be able to do more."



Click [here](#) to read her full blog post and visit NTSAD's Katie & Allie Buryk Research Fund [here](#).

First step is the Letters of Intent (LOI) and they are due no later than 8:00pm on Friday, September 16, 2016.

These grants are open to the international community (not limited to UPenn and CHOP). *Only academic institutions and nonprofit organizations are eligible.*

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