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ntsad research review

friday, september 19, 2014



Fourth Annual Day of Hope Benefits Research

The **Fourth Annual Day of Hope** is officially on **Saturday, September 20th** but families worldwide have been busy since early August and have events planned through October 24th. The funds raised will go directly to research further boosting the impact **NTSAD's Research Initiative** can have on current and future research projects. Since 2011, over \$90,000 in total has been raised for **Day of Hope** and with this year's successes we hope to reach \$130,000 in the next couple of months! *Your donation today could push us over that goal!*

Click [here](#) to see if any events are happening near you, or make a gift if you would like to support our efforts!

DONATE NOW
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Network for Good



The Abraham, Manning and Kenny Families at last year's Take a Walk for Tay-Sachs event near Philadelphia!

What is the NTSAD Research Initiative?

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Good Luck to Families Hosting Day of Hope Events This Weekend!

Your dedication and hard work will make such a difference. The impact you have on research will be significant!

Wishing the following families the best of weekends...

[The Benson Family](#)

in memory of Miss Elliott
(Badger Mountain, WA)

[The CATS Foundation and their Day of Hope Walk](#)

(Cardiff, UK)

[The Davis Boys](#)

in memory of Adam
(Portland, OR)

[The DuVall Family](#)

in memory of Nickolas
(Audubon, IA)

[The Manning Family in memory of Dylan \(DJ\)](#)

(Dyer's Deli, Parkesburg, PA and Brockton, MA)

NTSAD has funded research since its beginnings in the 1950s. It led to the discovery of the Tay-Sachs gene and the subsequent advent of carrier screening. In 2002, the Research Initiative was formalized and began funding cutting-edge research and collaborating with other rare disease groups to make the most of the grants. **Over \$3 million has been raised cumulatively for research since the Research Initiative began in 2002.** The onset of advancing technologies took research to a new level and the formation of the Tay-Sachs Gene Therapy Consortium in 2007 changed the landscape of NTSAD's research portfolio.

Now, the future of the Research Initiative will encompass not only basic and translational research, but it will also focus on clinical trial readiness as the possibility of new therapies shine on the horizon. One component of clinical trial readiness are natural history studies. A natural history study gives researchers a clear picture of how a disease advances and how to measure the effectiveness of a therapy. The article below summarizes the natural history studies of our family of diseases conducted to date, much of which has been funded by NTSAD.

To read more about the Research Initiative, click [here](#) to NTSAD's website.

Natural History Studies: An Important Part of Getting Ready for Clinical Trials

Written by Allison Bradbury, PhD and Staci Kallish, DO

Data collected from natural history studies is garnering importance as success in research moves us closer to initiating clinical trials. It is vital to have a baseline knowledge of the disease progression from which to measure the success of therapy. Researchers are conducting these vital natural history studies and collecting fundamental data to inform clinical trials. Furthermore, natural history studies provide valuable information to families and clinicians to help manage these difficult diseases. Natural history work is ongoing for many of our diseases with the aim of preparing to fund and conduct more natural history studies in preparation for future clinical trials. **NTSAD, to date, has made over \$354,000 in grants for natural history studies and has helped in gathering essential data for the studies.**

In 2011, Dr. Florian Eichler's group, at Massachusetts General Hospital in Boston, published an unprecedented natural history study on infantile GM2 gangliosidosis ([Read Pediatrics 128: e1233-1241 here.](#))

- The study included data on symptom onset and

[The Ohle-Rodriguez/Artinian Family in honor of Emma](#)
(Fredericksburg, VA)

[The Pounds Family in honor of Annabel](#)
(Murfreesburg, TN)

[The Prasad Family in memory of Abhinav](#)

[The Urban Family](#)
in memory of Brookie
(Bay City, MI)

Thank you to all who have supported the **Day of Hope** and to those who make gifts to make a difference!

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***Tip of the day from NTSAD parent and board member Brian Manning:***

Check with your company's matching gift program to leverage what you raise for research. You could very well double what goes to research!

**NTSAD featured in the September 2014 issue of**

**Indications™**  
Newsletter of The Lysosomal Disease Network™

The Lysosomal Disease Network features a patient group each month that is a part of the network, and NTSAD is the feature group this month. We thank them for highlighting us and look forward to future collaborations!

Read the September issue of "Indications" [here](#).

**Support the Sheep  
Support Hope**

disease progression of 92 patients with infantile GM2 (Sandhoff and Tay-Sachs disease) that were surveyed through NTSAD.

- The mean age at onset of earliest symptom was  $5.0 \pm 3.3$  months and the average age at diagnosis was  $13.3 \pm 5.3$  months. The most common initial symptoms were developmental arrest (83%), abnormal startle response (65%), and low muscle tone (60%).
- 98% of patients had seizures and the majority required multiple seizure medications, 75% of patients were partially or completely fed by G tube, and 88% required regular suctioning.
- Based on this study the researchers were able to develop a clinical severity scoring system based on the time at which milestones are lost and new symptoms arise.

**Dr. Eichler's group also completed a natural history study further characterizing juvenile and adult-onset types of Late Onset Tay-Sachs (LOTS).**

- Retrospective data on symptom onset and progression was acquired from 55 patients that were surveyed through NTSAD.
- The most common initial symptoms were the same in juvenile and adult LOTS and included difficulty running, speech abnormalities, impaired gross motor skills, inability to climb stairs, and impaired fine motor skills.
- The median time of onset of symptoms to becoming wheelchair-bound was 4 years in juvenile patients and 26 years in adult patients. Similarly, juvenile patients lost the ability to climb the stairs in the 1st decade of life, while adult patients lost this ability in the 3rd or 4th decade of life.
- Further defining the disease progression between juvenile and adult-onset LOTS will provide important information for patients, their families, and clinicians in managing and tracking this disease.

**Dr. Cynthia Tifft, at NIH, has conducted a natural history study on patients with GM1 gangliosidosis.**

- Repeated MRI scans of 5 late infantile GM1 patients showed rapid and similar progression from the first scan (median 6.5 months from onset of symptoms) to the second scan (median 43 months from onset of symptoms).



The Jacob Sheep continue to have an impact on the [Tay-Sachs Gene Therapy research](#). A part of keeping the flock means caring for them and making sure their pastures stay green. At least \$27,000 needs to be raised in the next six to nine months to continue the daily maintenance of the sheep.

**We need your help.**

In addition to gifts, there are new lambs you can adopt and name for a minimum gift of \$1,000. Contact Joan Lawrence at [joan@ntsad.org](mailto:joan@ntsad.org) for more information.

**Read more about current and past grants funded by NTSAD on our website [here](#).**



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- In contrast, serial MRI evaluation of 10 juvenile GM1 patients did not show significant changes in the severity of the MRI score over the course of the study.
- Dr. Tiff is also conducting magnetic resonance spectroscopy (MRS), which uses another type of imaging. She has found alterations of brain metabolites such as NAA, a molecule present in neurons, in GM1 patients.
- Alterations of MRI and brain metabolites determined by MRS could provide a non-invasive manner to track disease progression in affected children and measure therapeutic efficacy in clinical trials.

**A natural history study for the gangliosidoses is also currently underway at the University of Minnesota led by Dr. Chester Whitley.** It is a natural history study in which researchers hope to collect multiple sets of information from patients over time. They hope to include 15 Tay-Sachs disease/Sandhoff disease and 15 LOTS disease patients. This study will help researchers to better understand disease progression and continue to develop objective measures of disease severity.

**Dr. Paola Leone was a principal author on a 2006 Canavan Disease natural history publication** (Neuropediatrics 2006; 37: 209-221). This study was designed to model the natural history of Canavan disease using MRI and proton magnetic resonance spectroscopy (1H-MRS - another type of imaging). Researchers were able to demonstrate elevation of nerve specific brain metabolite, NAA, and glial cell (another important type of cell in the brain), specific metabolite, myoinositol. This study also used MRI to quantify changes in myelin formation and brain atrophy in specific regions of the brain. There is interest in building on this research to be able to monitor future studies involving drug, gene therapy, or stem cell treatments.

more.

**Stay Connected  
to NTSAD!**



I have become my own version of an optimist. If I can't make it through one door, I'll go through another door, or I'll make a door. Something terrific will come no matter how dark the present."

*- Rabindranath Tagore, Indian writer*

**national tay-sachs & allied diseases association**

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