



National Tay-Sachs & Allied  
Diseases Association, Inc.

Friday, AUGUST 14, 2015

## NTSAD Monthly Research Review

### Studies Essential to Research

#### The Importance of Natural History Studies

**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 against 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 funding more natural history studies in preparation for future clinical trials.

**NTSAD, to date, has made over \$478,000 in grants for natural history studies and has helped in gathering essential data for the studies.**

Read more about the studies funded that are focused on natural history [here](#).

Note: Last year's recipient of the Million Dollar Bike Ride grant was Annette Bley, MD, of Hamburg, Germany who is creating a natural history study focused on Canavan disease. She will be working with Drs. Florian Eichler and Heather Lau on this project as well. Read more about her work [here](#).

Read [here](#) about OpenApp that is developing a European rare disease registry focused on Tay-Sachs and Sandhoff

### Grant Opportunity!



One \$43,000 pilot grant is available focusing on forms of Tay-Sachs, Sandhoff, GM-1, or Canavan disease. We are soliciting proposals for innovative research projects that involve basic research, translational studies or clinical studies relevant to the diseases mentioned above.

Projects may be focused on:

(1) technology approaches such as stem cells, molecular chaperones, substrate inhibitors, small molecule drug screening, gene therapy, novel drug

*diseases with the help of the Cure & Action for Tay-Sachs (CATS) and Acción y Cura para Tay-Sachs (ACTAY) foundations.*

*NTSAD is currently working with researchers to determine what data should be collected directly from patients in addition to data derived from the clinical-driven studies that are funded by NTSAD. Stay tuned.*

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## Late Onset Focus Group and Assessment Update

Sue Kahn, Executive Director, and Diana Pagonis, Director of Family Services, recently met with the team who conducted the Late Onset focus groups and neurological assessments at the 2015 Annual Family Conference in Reston, Virginia. Gerry Cox, MD, Florian Eichler, MD, Alaa Hamed, MD, Julie Kissell, CGC, Swati Sathe, MD, and Cynthia Tifft, MD, PhD, were involved in these focus groups and neurological assessments to gain a better understanding of the progression and effects of Late Onset Tay-Sachs and Late Onset Sandhoff diseases.

At the meeting, Dr. Cox and Dr. Hamed shared summaries of the data analyzed that were collected in those focus groups. The assessments focused on how the disease affects patients from the physical side (gait, dexterity, speech). The data and measurements can be used effectively for a trial as they correlate well with measures of disease progression. At the conclusion of the meeting, the team of clinician researchers suggested meeting every other week to make progress on the next steps involved in understanding natural history and clarifying clinical outcome measures.

One take-away was how important it is for researchers, pharmaceutical companies and the FDA to understand a rare genetic disease, the number of individuals and families affected worldwide, as well as its progression and its impact on an individual/family in order to move forward with developing a therapy.

delivery to the brain or on

(2) other pre-clinical and clinical research needs, such as clinical outcome measures, registries, animal models, or biomarkers.

This grant is made possible by Team NTSAD, the National Tay-Sachs & Allied Diseases Association, and the Orphan Disease Center at PennMedicine.

[Read the announcement here.](#)

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## Mark Your Calendars!



**Next year's  
Third Annual Million Dollar  
Bike Ride will take place on  
May 7, 2016 in Philadelphia.**

Read the announcement [here](#).

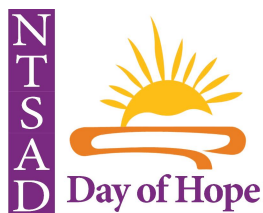


If you are an individual/family affected by Tay-Sachs, Canavan, GM1 or Sandhoff disease in any of their forms (infantile, juvenile, or late onset) and haven't connected formally with NTSAD, please [download this form here](#) and email it to Diana Pangonis at [diana@ntsad.org](mailto:diana@ntsad.org).

Being part of this process is being a part of research that could very well lead to an effective therapy for these diseases!

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## 2015 Fifth Annual Day of Hope



Families and friends of NTSAD are in the midst of planning their activities for this year's Day of Hope!

Our goal is to raise at least \$50,000 which could provide at least one researcher with the tools to advance their work from "bench to bedside!" Let Diana at [diana@ntsad.org](mailto:diana@ntsad.org) know if you want to organize something to commemorate the day - no idea is too small or too big!

[Visit this page](#) often to see if an event is happening near you. All proceeds benefit RESEARCH to give HOPE to our rare disease community worldwide. Hope Knows No Boundaries!

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Difference Tomorrow**

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