



National Tay-Sachs & Allied
Diseases Association, Inc.

Friday,
DECEMBER
16, 2016

NTSAD
Monthly
Research
Review

RESEARCH PROGRESS UPDATE

Tay-Sachs Gene Therapy Consortium

The **Tay-Sachs Gene Therapy Consortium**, led by Miguel Sena Esteves, PhD at the University of Massachusetts Medical Center, is carrying out IND-enabling studies, which are the last steps prior to filing an IND (Investigational New Drug) application and starting clinical trials.

We are pleased to report that the short-term safety study in normal Non-Human Primates (NHPs), which was initiated in September, has been successfully completed! Further pathology studies will be conducted, but the initial examination of the brain showed no appreciable changes. Some of the NHPs will be followed for up to one year. The grant for these studies was funded by the Cure Tay-Sachs Foundation.

In parallel, efficacy and safety studies in Sandhoff mice are being conducted. The first GM2 mice were injected in September and the last two mice are expected to be injected soon. This study requires 90 mice (75 GM2 and 15 normal). Based on the 5-month of age endpoint, the animal study is expected to be completed by the end of April 2017 with biochemical studies completed in all tissues by September 2017. This grant for these studies is being funded by NTSAD.

Other components of the IND-enabling studies are carrying out GLP-toxicology and biodistribution studies in certified labs as well as starting production of manufacturing vectors for the clinical trial. Funding for these activities is still needed.



IN THIS ISSUE

[Tay-Sachs Gene Therapy: One Step Closer](#)

[Subcommittee on Experimental Therapies](#)

[Ketogenic Diet Review](#)

[Use of Medical Marijuana for Seizures](#)

[NTSAD Request for Research Proposals](#)

[Breakthrough Act Signed into Law](#)

[Canavan Patient Insights Network](#)

NTSAD SCIENTIFIC ADVISORY COMMITTEE'S

SUBCOMMITTEE ON EXPERIMENTAL THERAPIES

The goal of the subcommittee is to evaluate clinical trials or other proposed therapies for Tay-Sachs, GM1, Sandhoff, Canavan and related diseases to educate families regarding trials and how to assess a proposed new or untested therapy. Following are two papers written by two of NTSAD's Scientific Advisory Committee members that address the ketogenic diet and the medical use of marijuana to control seizures which are topics that have been discussed within our community.

Could Ketogenic Diets Become a Complimentary Therapy Strategy for Ganglioside Storage Diseases?

Thomas Seyfried, PhD, Biology Department, Boston College and Christine Denny, PhD, Department of Psychiatry, Columbia University Medical Center

Drs. Seyfried and Denny review an experimental therapy about how diet can impact inflammation in chronic inflammatory diseases by explaining the ketogenic diet, its potential effects and how it can supplement and/or complement a substrate reduction therapy.

Read the paper in its entirety [here](#).

Medical Marijuana in Epilepsy

Swati Sathe, MD, Director, Pompe Clinical Development, Amicus Therapeutics

21st Century Cures Act:

Signed into Law

President Obama signed the 21st Century Cures Act into law this week after the Senate voted 94-5 in favor of passing this important legislation that could impact the lives of many in the rare disease community.



Canavan Patient Insights Network - Join Today

Max's mom, Ilyce Randell, has worked tirelessly to advocate for her son, Max, who has Canavan disease, and the many other children who are affected by Canavan disease.



Through her foundation, Canavan Research Illinois, she recently initiated a type of registry called a Patient Insights Network (PIN) through the company AltaVoice. To date, 90 Canavan families have entered their information to join this PIN. She has invited NTSAD to collaborate in the Canavan PIN with a goal of bringing together the Canavan patient community. She has also invited Sue Kahn, our Executive Director, to join the Canavan PIN Advisory Committee (CPAC) as the representative from NTSAD. [Here is the link](#) if you would like to sign up. If you have questions, please contact NTSAD at (617) 277-4463.

Dr. Sathe reviews the information available regarding the medical use of marijuana to treat epilepsy. Because not all medications may treat seizures effectively, some families turn to other modalities of treatment to control them in their loved ones. This paper is a helpful outline regarding the use of medical marijuana (THC, CBD) for seizure management. As always, health professionals should be consulted if alternative, natural or experimental therapies are being explored.

Read the paper in its entirety [here](#).

Did you know the **CARERS Act (S.683)** was introduced to the Senate by Senators Corey Booker (D-NJ), Rand Paul (R-KY), and Kirsten Gillibrand (D-NY) and in the House by Representatives Steve Cohen (D-TN) and Don Young (R-AK), and that this bipartisan legislation would remedy the state-federal conflict over medical marijuana laws?



Read the full bill [here](#) and what it means [here](#).

NTSAD's REQUEST FOR PROPOSALS

Pre-application deadline is January 4, 2017

The **NTSAD Research Initiative** funds research that may lead to treatments for lysosomal storage diseases or leukodystrophies impacting the central nervous system (CNS). NTSAD is now soliciting proposals for innovative research projects that involve basic research, translational studies or clinical studies in the following diseases: Tay-Sachs, GM-1, Sandhoff, and Canavan disease). Basic research and translational studies should generate strong preliminary data to enable future major funding by other third parties. Projects may be in such areas as drug delivery to the brain, new animal models designed to facilitate translational research and drug discovery, assay development for drug screening, substrate reduction, stem cells, molecular chaperones, gene therapy, and biomarkers, as well as exploring other novel therapeutic strategies.

All proposals are reviewed and evaluated by members of the NTSAD Scientific Advisory Committee and other qualified scientists. The final decision of award

The **AltaVoice Canavan PIN** is separate from the current natural history study conducted by Heather Lau, MD at NYU, Principal Investigator and her co-investigator, Paola Leone, PhD. This study is co-funded by NTSAD and the Canavan Foundation. Both of these efforts contribute to the clinical trial readiness for future trials that may benefit Canavan disease.

NTSAD is currently evaluating options to establish a formal registry for the gangliosidoses. Stay tuned for future news on this topic.

Make a Gift [HERE](#)
for Research



Are you on Facebook and Twitter?



Like **NTSAD's Facebook page** and follow **@susanrkahn** for new developments in therapies, science, genetic testing, patient advocacy, and more.

recipients is determined by the NTSAD Board of Directors.

Download the application guidelines [here](#).

Timelines:

Pre-application deadline:

5:00pm (EST), Wednesday, January 4, 2017

Invitation to submit a full application no later than:

Monday, January 30, 2017

Full application deadline (by invitation only):

Wednesday, February 22, 2017

Earliest anticipated start date: Wednesday, July 1, 2017



national tay-sachs & allied diseases association

| | |
|---|--|
| susan kahn, executive director (skahn@ntsad.org) | 2001 beacon street |
| joan lawrence, development director (joan@ntsad.org) | suite 204 |
| diana pangonis, family services director (diana@ntsad.org) | boston, ma 02135 |
| ingrid miller, office manager (ingrid@ntsad.org) | (617) 277-4463 |
| becky benson, conference coordinator (becky@ntsad.org) | |
| veronica huang, science communications & outreach intern (veronica@ntsad.org) | |
| emily randall, development associate (emily@ntsad.org) | www.NTSAD.org |

