



National Tay-Sachs & Allied
Diseases Association, Inc.

Friday, FEBRUARY 15, 2016

NTSAD Monthly Research Review

New Research Grant Funded!

Vera's Research Fund of NTSAD, the Heringer Fund for Research of NTSAD, and The LOTS Research & Education Foundation are together funding a new project to identify Tay-Sachs disease biomarkers using lipidomics technology.

In a promising area of biomarker development called lipidomics, many different lipids are analyzed in a given sample. The primary storage product of Tay-Sachs disease, GM2 ganglioside, is a lipid. This research project will use the sheep model of Tay-Sachs disease to identify biomarkers which could then be validated in patient samples.

The Co-Principal Investigators are Daniel S. Ory, MD, from Washington University in St. Louis, and Doug Martin, PhD, from Auburn University College of Veterinary Medicine.

MRI and MRS as a surrogate marker for clinical progression in GM1 Gangliosidosis

Dr. Cynthia Tiffet and her group from the NIH published an article in the American Journal of Medical Genetics in December 2015 discussing the use of imaging techniques to monitor progression of disease in patients with GM1 gangliosidosis.

They used MRI (magnetic resonance imaging) and MRS

Jacob Sheep: A Flock Helping Research



The Jacob Sheep flock needs your help! There are currently over 20 pregnant ewes and some with twins so in the next 2 months, there will be 20-30 new lambs born, and lots of naming opportunities!

If you would like to help fund research and help maintain the sheep flock by naming a sheep for \$1,000 or participate in the Adopt-A-Sheep program, please contact the office [here](#).

There is also yarn available at \$20 per skein made from the Jacob's Sheep wool. Contact Ingrid [here](#) for more information.

The summary of the three year sheep study is available [here](#) in our Library.

(magnetic resonance spectroscopy) imaging combined with scoring systems to measure motor function and language skills in 6 children with infantile GM1 and 9 with juvenile GM1. They found the measures of brain volume on MRI and markers on MRS correlated with the disease progression in patients. As expected, those with infantile GM1 showed more significant abnormalities on imaging with faster progression when compared to those with juvenile GM1. This suggests that MRI and MRS could be used for clinical outcome measures in future clinical trials.

What are Biomarkers?

Why are they so important?

Biomarkers are critical for understanding disease progression. Like natural history studies, biomarkers are an important part of clinical trial readiness.

Often biomarkers, such as compounds measured in blood, urine, or cerebrospinal fluid, can show improvements after therapy faster than clinical improvement can be seen in a patient. Knowing the typical progression of a disease using natural history data, chemical biomarkers, or imaging findings is crucial to be able to show that an experimental treatment is improving the disease course.

2015-2016 Research Initiative Request for Proposals: Next Step - Full Applications

NTSAD's Research Chairs, Drs. Fran Platt (SAC) and Staci Kallish (Research Initiative Committee), recently reviewed the scores and ranking for the one page pre-applications. It was a difficult decision, but 10 of 19 applicants are now invited to now submit a full application.

This two-step process is similar to that of many other medical research foundations. The grant topics were varied and of interest: identifying new small molecules for treatments, evaluating new biomarkers, new methods of drug delivery to cross the blood brain barrier, as well as hot new research topics such as CRISPR.

Presented by Dr. Cynthia Tjff



Save the Date for a Special
**GM-1
GANGLIOSIDOSIS
WORKSHOP**

Saturday, April 9th, 2016
NTSAD Conference
Orlando, FL USA

A Rare Disease Collaboration



Sponsored by
LYSOGENE

To attend this workshop and/or the Annual Family Conference, contact **Diana** or **Becky** for more info.

THIRD ANNUAL

MILLION DOLLAR BIKE RIDE



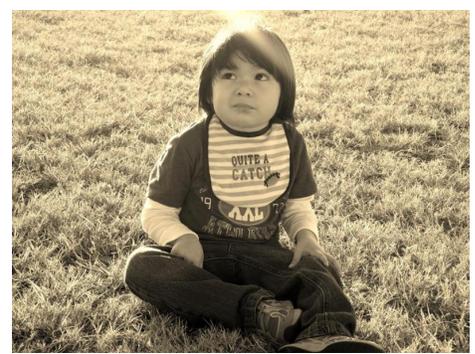
Ride with Team NTSAD Saturday, May 7, 2016

If we raise at least \$20,000, these gifts will be matched up to \$50,000 to fund a research grant focused on our group of rare genetic diseases.

If you're interested in riding with Team NTSAD in Philadelphia, contact Allison Bradbury [here](#).

The 2015 Team NTSAD MDBR grant award went to Marlene Jacobson, PhD, of Temple U. for her project, "Patient-Derived Phenotypic Assay to Discover Treatments for Tay-Sachs Disease". The **2014 MDBR grant award went to Annette Bley, MD**, University Hospital - Hamburg, for "Quantitative description of the clinical course of Canavan disease".

The pre-applications were ranked scientifically and strategically, i.e., relevance to our **Request for Proposal (RFP)**. The top two criteria were strong science and fit with Clinical Trial Readiness. The **Scientific Advisory Committee (SAC)** ranked the pre-applications on both dimensions whereas the **Corporate Advisory Council** ranked them on strategic interest.



We wish we had more funds to expand our grant funding for the grants received in response to the RFP. We are **currently funding** the Tay-Sachs Gene Therapy Consortium in addition to six Research Initiative grants that have been awarded since 2014. Because of these funding commitments, it's likely that we'll be able to fund just two, or possibly three, of the new grant applications. If our uncommitted funds increase, we will be happy to award more high quality grants.

Make a Gift to Support Research [here](#) today.

Are you on Facebook and Twitter?



Like **NTSAD's Facebook page** and follow [@susankahn](#) for new developments in therapies, science, genetic testing, patient advocacy, and more.

WORLD Meeting * San Diego, California February 29th to March 4, 2016

The 12th annual **WORLD** (We're Organizing Research for Lysosomal Diseases) meeting is coming up in a few weeks and a number of Scientific Advisory Committee (SAC), Corporate Advisory Council (CAC), and NTSAD Board members will be in attendance.

Please contact NTSAD's Executive Director, Sue Kahn, if you'd like to connect with NTSAD or their expert advisers during WORLD. She has already organized several meetings with companies and researchers.

The program includes one presentation each day about our diseases. Stay tuned for the March Research Review because our research communications team, Allison Bradbury and Staci Kallish, will share the WORLD highlights with you and what it means for advancing research.

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