National Tay-Sachs & Allied Diseases Association, Inc.

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NTSAD Monthly Research Review

Interim Progress Report: "Development and Validation of an MS-MS Method for the Detection of Hexosaminidase Deficiency in Tay-Sachs Disease."

Denis C. Lehotay, PhD, Principal Investigator
Queen's University, Kingston, Ontario

Description and Status: The goal of the project is to develop a screening test for newborns with Tay-Sachs disease (TSD) using a tandem mass spectrometry method. The 12-month project report noted that (1) researchers are currently mapping the baseline activity of hexosaminidase in normal patients, (2) they are working out a protocol to allow testing in a large number of patient samples, and (3) their initial experiments indicate that the assay works well to measure both normal patients and those affected with Tay-Sachs disease.

Impact: Once treatment for TSD becomes available, detecting TSD by screening and initiating early treatment will become essential. Developing, validating, and testing a rapid assay for measuring hexosaminidase activity in populations with a high incidence of the disease are part of the essential steps that will eventually lead to a cure. Tandem mass spectrometry has been clinically studied to establish cutoff ranges of biomarkers for rare metabolic disorders in newborn screening [1]. Such data and information will be highly beneficial for the screening of Tay-Sachs disease. The ongoing project will generate relevant data (reference ranges and cutoff values of key biomarkers) in the population of French Canadians which is a group with a high carrier frequency. Ultimately the hope is to use this assay for newborn screening in a general population.

Read more about the project here.

[1] McHugh, DMS, etc. Clinical validation of cutoff target ranges in newborn screening of metabolic disorders by tandem mass spectrometry: A worldwide collaborative project, Genetics in Medicine, 2011, 13, 230-254.

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THANK YOU TO...

NTSAD would like to thank James F. Ewing, JD, PhD and Jolene S. Fernandes, JD, PhD, Attorneys at Foley & Lardner, LLP in Boston. Both Jim and Jolene have an expertise in intellectual property law. We are fortunate that they have been advising NTSAD on a pro bono basis in recent months on language for our grant agreements. A former attorney with Foley & Lardner also filed a patent application based on NTSAD-funded research. While the patent did not issue, we will continue to work with them to seek similar opportunities.

INTRAVENOUS GENE THERAPY IN TAY-SACHS SHEEP

Heather Gray-Edwards, PhD, Auburn University

Heather Gray-Edwards, PhD, from Auburn University received an NIH K08 grant to study intravenous gene therapy in the Tay-Sachs sheep. This study will test novel AAV constructs and new AAV capsids to increase efficiency of gene delivery and avoid invasive brain injections. Sheep will be injected intravenously at an early age and clinical disease will be evaluated using biomarkers including neurologic examination, advanced ultra-high field MRI and targeted lipidomics. This research complements other gene therapy projects in the Sandhoff mice and cats because the Tay-Sachs sheep are the only Tay-Sachs model in which the clinical effects of gene therapy can be evaluated (Tay-Sachs mice have no clinical symptoms). Since the sheep brain is much larger, approximately ¼ the size of a human brain, this study will provide information on methods to overcome challenges associated with increased brain size. This research will be done in collaboration with Daniel Ory, PhD, Miguel Sena-Esteves, PhD, and Douglas Martin, PhD. Since 2010, NTSAD has funded $330,000 in grants for related studies in this Jacob sheep model of Tay-Sachs disease, including $42,000 to support the care and feeding of the sheep flock. This continuous support of the Tay-Sachs sheep breeding flock enables studies like this new one to occur.

URGENT: NEED YOUR HELP

The **21st Century Cures Act** passed in the House in the summer of 2015, and it's been in the Senate since then. Unfortunately, if the bill is not passed now, it will die on the floor and the hard work of thousands of advocates will be for naught. However, with your help there is still a chance.

We heard from our friends at **EveryLife Foundation** that two senators need to hear from you NOW. Below are their names and a script that follows. It only takes a couple of minutes to make the call. Every call that the legislators receive must be logged in. We need to convince these senators that it isn't just about business, this bill represents real change for families across the country.

RESEARCH GRANTS

2017 Request for Proposals

Stay tuned to the December **Research Review** for the 2017 NTSAD Request for Proposal announcement.

The **NTSAD Research Initiative** funds research that may lead to treatments for lysosomal storage diseases or leukodystrophies impacting the central nervous system (CNS). NTSAD solicits proposals annually for innovative research projects that involve basic research, translational studies or clinical studies. Basic research and translational studies should generate strong preliminary data to enable future major funding.

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

Make a Gift HERE for Research

Are you on Facebook and Twitter?

Please. Help make a difference with just one phone call. Thank you.

Office of Senator Elizabeth Warren (D-MA) (202) 224-4543
Office of Senator Patty Murray (D-WA) (202) 224-2621

* My name is XXXX and I'm calling from [use your city and state if constituent]
* I'm calling today to urge the Senator support the 21st Century Cures Act and ensure that the OPEN ACT - Senate bill 1421 is included
* The OPEN ACT is life-saving legislation that could double the number of therapies available to rare disease patients
* This bipartisan legislation has the support of nearly 170 patient organizations across the country and they are counting on your office to stand up for patients who do not yet have an FDA-approved treatment
* Thank you for your work to advance health for patients across the nation

Thank you for your support. Together, we can get the OPEN ACT passed!

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

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