NTSAD Be Informed Webinar Series II

NTSAD broadcasted the second webinar in the NTSAD Webinar Series "Be Informed" on Friday, December 6th. The purpose of these webinars is to share information in an accessible and helpful way for families as we begin to navigate complicated topics.

NTSAD Board member Dr. Gerald Cox shared his expertise and knowledge in a presentation about clinical trials with the goal to demystify how they are structured, the purpose of a trial and the oversight that is required to ensure it is successful.

SAVE THE DATE: Thursday, January 23, 2020 for the next **NTSAD Be Informed** webinar that will review the regulatory process behind drug development presented by Curt Scribner, MD, an independent biotech regulatory consultant, who has worked for the FDA as well as a number of biotech and pharma companies.

A Visit to Aspa Therapeutics

NTSAD, Canavan Foundation and **Canavan Research Illinois** were invited to visit Aspa Therapeutics laboratory to get a tour, learn about development of their Canavan gene therapy program, and, most importantly, to share with them the perspectives of our organizations as we represent families.

The team explained the steps in producing the vector they're using for the gene therapy from making the actual virus to the final vector. The pictures below show some of the highlights of these steps. Another message that came across clearly was how important it is for families to participate in the CANinform Natural History Study. The data from these studies tells the story of Canavan disease progressoin and accompanies the hard work from the lab to make a compelling case for the FDA to approve clinical trials.

Thank you to Ilyce Randell for sharing these photos with NTSAD.

Canavan Families CANinform

By sharing information, Canavan families canhelp speed up clinical development of the gene therapy, which will benefit the Canavan community.

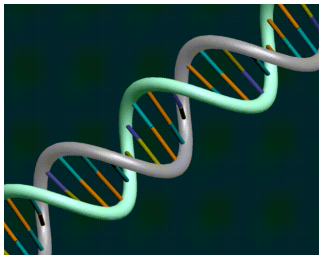
The study is open to all families with a confirmed diagnosis of Canavan disease. Medical records are a key component for gathering this data and are needed to participate in CANinform. Aspa is offering a record retrieval service to help families (outside of the EU) to obtain past medical records using three simple steps, detailed below. Visit **treatcanavan.com** for more information.

CALL - 1 (833) 764-2267 or email **CanavanMedRec@veristat.com** to use a free service to collect records.

Then **CONSENT** - Fill out a short online form to provide permission to share medical information and begin the process of obtaining records.

Lastly, **CONTRIBUTE** - Once records are retrieved, enroll your child in CANinform, the Canavan Natural History Study.

UPDATED:
NTSAD's Tay-Sachs Carrier Screening Position Statement



In April, 2019, NTSAD's **Scientific Advisory Committee (SAC)** formed a Task Force led by Dr. Jodi Hoffman, with fellow SAC members Marvin Natowicz, MD, PhD, Mimi Blitzer, PhD and her U. of Maryland colleague, Erin Strovel, PhD, and board members Karen Grinzaid, MS, CGC and Gerry Cox, MD, PhD. The purpose of this task force was to create a position statement regarding the use of next-generation sequencing (NGS) of the HEXA gene compared to other technologies for Tay-Sachs carrier screening. Also, the purpose was to update NTSAD's last position statement which was completed in August 2009.

The conclusion is as follows: Two recent studies* suggest equal or better performance characteristics of full exon sequencing by NGS of the HEXA gene compared to Hex A enzyme activity testing for Tay-Sachs carrier screening (Hoffman et al, 2013; Cecchi et al, 2019). Although future studies may provide further data, the results of these studies suggest shifting toward routine use of full exon sequencing of HEXA for Tay-Sachs carrier screening in individuals of all ethnic backgrounds due to the benefits and few limitations of NGS, while continuing to regard Hex A enzyme activity testing as another reliable method for Tay-Sachs carrier status detection.

This statement is being shared with the relevant medical associations, i.e., American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics and Genomics (ACMG), along with the testing laboratories. In January, we will share more information about carrier screening and the Tay-Sachs Carrier Screening Position statement.

**One of these studies was funded by NTSAD, Cameron and Hayden Lord Foundation, Mathew Forbes Romer Foundation, Evan Lee Ungerleider Fund of NTSAD and the NTSAD New York Area Fund.*

IntraBio Clinical Trial: Update

IntraBio Ltd. is pleased to share that the IB1001-202 clinical trial has screened nearly 50% the target number of patients.

The study, which investigates N-acetyl-L-leucine (IB1001) for the treatment of GM2 Gangliosidosis (Tay-Sachs and Sandhoff) will enroll a total of approximately 30 patients across all international sites. Since recruitment commenced, fourteen (14) patients have been screened across eight (8) international clinical trial sites.

Recruitment is expected to continue rapidly and be completed by Spring 2020.

In the US, patients aged 18 and older with a confirmed genetic diagnosis of Tay-Sachs of Sandhoff disease may be eligible for recruitment. The trial will consist of three study phases: a baseline period, a 6-week treatment period, and a 6-week post-treatment washout period. A planned one-year extension phase has recently been approved in Europe. Patients (and a caregiver) will be reimbursed for reasonable out-of-pocket expenses incurred for participating in the trial, such as travel and parking.

To ensure all patients have the opportunity to participate, interested patients are encouraged to contact their nearest study center as soon as possible to schedule a screening visit before enrollment is complete.
Information on each site, including contact details, can be found below.

NYU Langone School of Medicine
New York, New York, United States, 10017
Principal Investigator: Dr Heather Lau, MD Contact: Sara Rodriguez, Research Coordinator Email **here**.
Tel: 929-455-5108

University of California - Los Angeles Los Angeles, California, United States, 90095
Principal Investigator: Dr Susan Perlman, MD Contact: Aaron Fisher, Staff Research Associate Supervisor Email **here**.
Tel: 310-206-8153

The Mayo Clinic
Rochester, Minnesota, United States, 55905
Principal Investigator: Dr Anhar Hassan, MD Contact: Sandra Looney, Clinical Research Coordinator Email **here**.
Tel: 507-538-4107

For the complete enrollment criteria, as well as details regarding the study assessments, multinational clinical trial sites, etc., please visit ClinicalTrials.Gov(NCT03759665) **here**.

Tay-Sachs in the 21st Century Conference

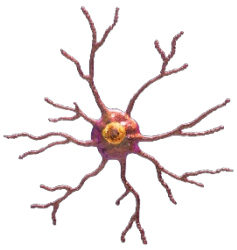
NTSAD Scientific Advisory Committee member, Steve Walkley, MD, DVM, organized the 7th Annual Isabelle Rapin Conference at Albert Einstein School of Medicine on December 4th in New York. In attendance were professionals, industry and members of the NTSAD family.

The speakers included many experts in the field of Tay-Sachs and the gangliosidoses. Their presentations ranged from the historical perspective (e.g., the 1887 publication first characterizing Tay-Sachs disease) to existing clinical trials to early research using CRISPR/Cas9-mediated DNA Base Editing for correcting mutations in Tay-Sachs disease. Bringing the family perspective to the meeting, Tim Lord, past NTSAD board president, and Alexis, Katie and Allie Buryk, shared their personal experiences and how Tay-Sachs has impacted their families.

Photos and a full program video will be available in January 2020.

NTSAD Research Grant Progress Report

Role of microglia in Sandhoff disease pathology
Tony Futerman, PhD
Weitzman Institute of Science, Israel

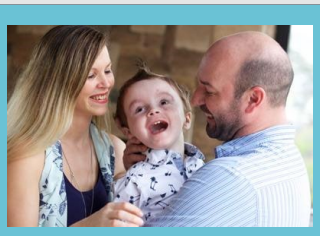


Sandhoff disease (a form of Tay-Sachs disease) is characterized by activation of immune cells in the brain in a process known as 'neuroinflammation'. The main immune cells in the brain are microglia, and in the current research project, the Futerman laboratory in the Weizmann Institute is isolating microglia from the brains of a mouse model of Sandhoff disease to characterize how these microglia change during disease progression. Currently, microglia have been isolated, and using a technique known as 'RNAseq', gene expression profiles have been determined from Sandhoff disease mice at a number of different stages of disease progression. The results suggest some unique patterns of microglia activation in Sandhoff disease, and some patterns which are similar to other lysosomal storage diseases. The goal is that by the end of the project some of these pathways might be options for novel therapeutic approaches.

Watch a video **here** to understand what microglia are and how they function.

Research is one way NTSAD supports families.

Make a year-end gift today to keep the wheels turning as we get closer to finding treatments.



Give Today.

NTSAD

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