

Canavan Update

Did you know there is a monthly phone call scheduled with a group of 18 clinicians, scientists and patient advocates who come from the U.S. and Germany? The group has grown as we connect to more people with an invested interest in the goals of the **Canavan Consortium**, which are conducting natural history studies and establishing a registry. Both are necessary for the success of any future clinical trials.

Read more about the natural history studies and Canavan Patient Insight Network below.



Happy 10th Birthday to Dylan (pictured right) and all those who honored his day with a gift to the [Canavan Research Fund](#)! Keep smiling!

Natural History Studies

NTSAD and the Canavan Foundation made a grant in 2015 to New York University for a project conducted by Heather Lau, MD and Paola Leone, PhD, of Rowan University, "Defining the Natural History of Canavan Disease through Development of an International Registry." Subsequently, Florian Eichler, MD at Massachusetts General Hospital (MGH) and Annette Bley, MD at the University Medical Center Hamburg-Eppendorf in Hamburg, Germany have joined. They all agreed to use the common NeuroBANK database which is housed at MGH. This study requires Institutional Review Board

AltaVoice:

Canavan Patient Insight Network

This initiative is intended to build a global patient community by reaching out to as many Canavan patients as possible and having them submit their information directly. This global Canavan Patient Insights Network (PIN) has located over patients thus far (40% from within the US, the rest international) by circulating information and a link to the registry through social media and email. It was started by Ilyce Randell who founded [Canavan Research Illinois](#) after

(IRB) approval and other documentation because it is being carried out in a rigorous way in a formal setting.

The purpose of this clinician-driven disease registry is to enhance the understanding of the natural history of Canavan disease. They are aiming to define appropriate clinical endpoints that may be used in the development of clinical trials for Canavan disease. The goal is for physicians to consistently share clinical data from their Canavan patients.

Who is eligible: Children & adults of any age with a confirmed diagnosis of Canavan disease

What will be required if you decide to participate:

- Read and sign a consent form
- Provide past medical records to the Study Team
- Complete a questionnaire about your child's development and symptoms related to Canavan disease
- Allow data collection from future doctor visits that are scheduled as part of your child's regular care

To enroll in the study, contact Rachel Duong, BA at (617) 724-1330 or

rtduong@partners.org.

her son, Max, was diagnosed with Canavan Disease. The Canavan Foundation and NTSAD are now also members of the Canavan PIN.

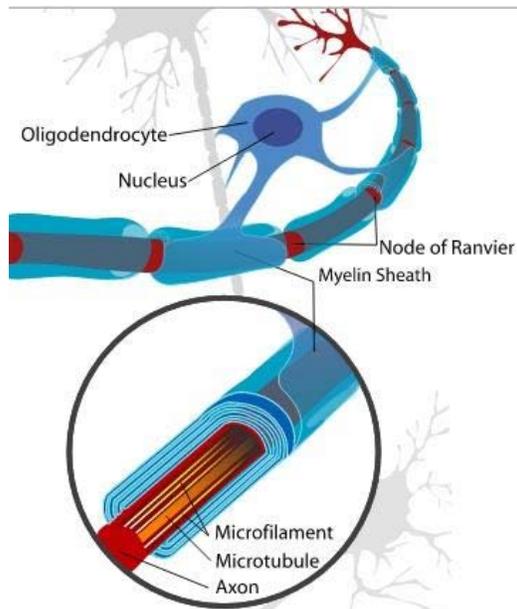
The platform consists of four general surveys and the capability to upload relevant medical records. It is easy to use and allows patients to maintain ownership of data. The platform allows families who register to set their communication settings to show whether they would like to be contacted with research opportunities. Currently 89% have indicated interest in communications about research. A Canavan specific survey is in development.

How will AltaVoice work with the researchers conducting the natural history studies?

An AltaVoice coordinator will send information about the natural history study to those who have indicated interest in research. This communication will contain contact information for the natural study staff so that patients can contact study coordinators if they would like more information.

The natural study sites will make participants aware of the AltaVoice Canavan PIN to help register as many patients as possible

WHAT IS CANAVAN DISEASE?



Canavan is a progressive neurological genetic disorder caused by the absence of a vital enzyme known as aspartoacylase (ASPA). ASPA breaks N-acetylaspartate acid (NAA) into building blocks essential for building myelin. Myelin is a fatty membrane (also known as white matter) that forms a protective coating around each nerve ensuring the nerve functions properly. It is one of over 40 genetically inherited disorders known as [Leukodystrophies](#).

Anyone can be a carrier of Canavan disease and not have any symptoms. When both parents are carriers, each child has a 25% of having the disease.

The carrier rate for the general population is 1/300. Ashkenazi Jews are at higher risk with a carrier rate of 1/55. Canavan disease is one of several genetic diseases along with Tay-Sachs included in the recommended Ashkenazi Jewish carrier screening panel.

(Learn more here about the other diseases in the panel [here](#).)



'You can't discount the potential ability of anybody': Boy with rare disease finally able to communicate

Richmond-Times Dispatch

By Katie O'Connor

He wears a visor on his head with a laser pointer peeking over the edge, its red light moving over the page of a letter board in front of him.

"You want to tell us something?" asks his physical therapist, Shannon LaSpina. "What do you want to tell us?" "Liam," his mother, Amy Heller, chimes in, "I want you to use complete sentences this time." It's not clear, at first, that Liam understands. He can only move his head incrementally with LaSpina's support, and he does not look at the letter board held up in front of him straight on. He sees out of the tops or sides of his eyes, instead, and his long, feathery lashes hide what he's seeing from the casual observer. Slowly, the laser pointer's red dot moves along the letter board. "I-C-A-N-T-A-L-K," Liam spells.

Read the full story [here](#) about Liam's ability to communicate despite having Canavan Disease.

Paola Leone, PhD, of Rowan University and featured in this story, is a long-time member of NTSAD's Scientific Advisory Committee.

SAVE THE DATE...SEPTEMBER 12TH

"Fore"



**The Ninth Annual
Jack Kliger Memorial Tennis & Golf Outing**



**THIS YEAR AT
Piping Rock Club
Locust Valley, NY
Tuesday, September 12, 2017**

Benefitting NTSAD's mission to lead the fight to treat and cure Tay-Sachs, Canavan and related genetic diseases and to support affected families and individuals in leading fuller lives.

Formal invitation to follow



Jack Alan Kliger was diagnosed with Canavan Disease when he was nine months old. Jack was given a life expectancy of three to five years. Jack passed away in April 2008 just shy of his 14th birthday.

Throughout his short life his family received support and encouragement by being part of the larger family of NTSAD and its members whose children fought similar diseases and similar battles.

The Kliger family invites you to join them in New York on September 12 to honor his life while giving back to NTSAD. Stay tuned to more information on how to register to golf or sponsor the event [here](#).

Wishing Karin Croft, Bonnie Pastor and Sophia Pesotchinsky best wishes for a successful event on Saturday, June 24th featuring the stylings of Shelley Komarov to benefit Late Onset research!

Gifts for Late Onset Research can be made to the Vera's Research Fund of NTSAD [here](#).

Susan Kahn, Executive Director
Brian Manning, President, NTSAD Board
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