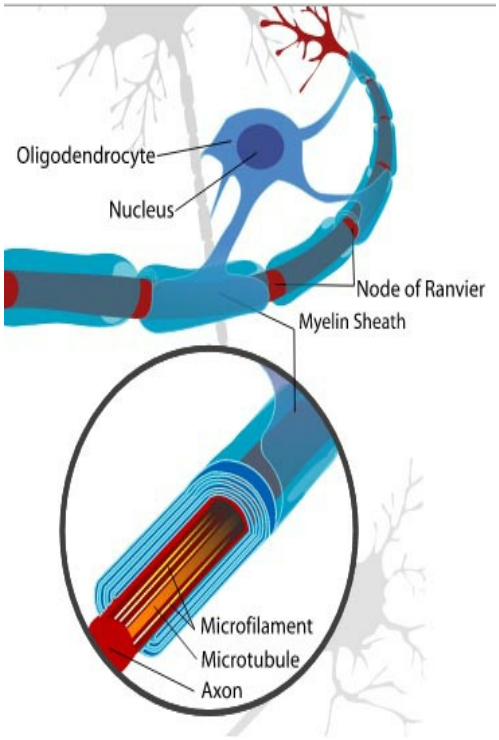
National Tay-Sachs & Allied Diseases Association, Inc.

Monday, SEPTEMBER 14, 2015

NTSAD Monthly Research Review



leukodystrophy:

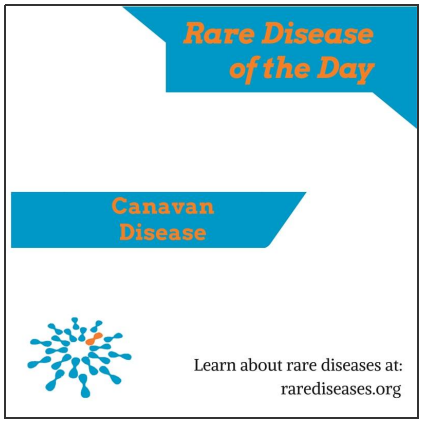
noun / leu·ko·dys·tro·phy (lōō'kō-dĭs'trə-fē)
leuko (white), dys (lack of) and troph (growth)

: a group of genetic disorders characterized by progressive degeneration of myelin (white matter) in the brain, spinal cord, and peripheral nerves.

: approximately 40 leukodystrophies including **Canavan** disease, adrenoleukodystrophy, Alexander's disease, Krabbe disease, metachromatic leukodystrophy, Sjogren-Larssen, and Zellweger syndrome.

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



Canavan was recently featured as

Pre-Application Deadline: September 14th

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**.

NORD's Rare Disease of the Day

What's Happening with Canavan Research?

NTSAD has funded over $250,000 in grants focused on Canavan disease over the last several years. They ranged from basic research, small molecules, gene therapy to clinical trial readiness.

Read about Dr. Guangping Gao's gene therapy work at the University of Massachusetts Medical Center in Worcester, MA.

- NTSAD Grant - Final Report **here**.
- **A Single Intravenous rAAV Injection as Late as P20 Achieves Efficacious and Sustained CNS Gene Therapy in Canavan Mice**, *Published in The American Society of Gene & Cell Therapy*

There are currently two NTSAD-funded projects focused on the natural history of Canavan disease. It is imperative to understand the progression of the disease to learn how to best measure the success of any clinical trial/treatment.

- *Defining the Natural History of Canavan Disease through Development of an International Registry,* Principal Investigator: Heather Lau, MD and Co-Investigator: Paola Leone, PhD (Co-funded with The Canavan Foundation)
- *Million Dollar Bike Ride Grant to Study Natural History of Canavan Disease,* Principal Investigator: Dr. Annette Bley, University Hospital of Hamburg Eppendorf, Germany

Read more here about Dr. Adeline Vanderver work with the Global Leukodystrophy Initiative (GLIA) and Dr. Florian Eichler's experience using the NeuroBANK for the ALD registry. They both presented at the NTSAD Science

One $43,000 pilot grant is available focusing on forms of Tay-Sachs, Sandhoff, GM-1, or Canavan disease. Seeking proposals for innovative research projects that involve basic research, translational studies or clinical studies relevant to the diseases mentioned above.

This grant is made possible by Team NTSAD, the National Tay-Sachs & Allied Diseases Association, and the Orphan Disease Center at PennMedicine.

See Guidelines **here**.

Pre-application deadline is September 14th. Apply **here**.

2015 Fifth Annual Day of Hope

Rally & Raise Hope about Tay-Sachs, GM1, Canavan, Sandhoff and related genetic diseases!

Visit this page to see if an event is happening near you. All proceeds benefit RESEARCH to give HOPE to our rare disease community worldwide.

Symposium & Workshop for Professionals held in Reston, Virginia in April 2015.

ALDConnect

Annual Meeting - Oct. 1-3, 2015

NTSAD Executive Director, Sue Kahn, was invited to talk about NTSAD and how NTSAD would like to collaborate with other Canavan and leukodystrophy organizations around the issues of understanding what's important to patients, better understanding disease, etc.

Too many children in the Canavan community gone too soon in the last year. Support Research **here**.

NTSAD Scientific Advisory Committee

Fran Platt, PhD (Chair)
Jodi Hoffman, MD (Vice chair)
Miriam Blitzer, PhD
Robert Desnick, PhD, MD
Florian Eichler, MD
Mark Haskins, VMD, MS, PhD
Michael Kaback, MD, FACMG
Edwin Kolodny, MD
Heather Lau, MD
Paola Leone, PhD
Heather Lau, MD
Paola Leone, PhDGustavo Maegawa, MD, PhD
Marvin Natowicz, MD, PhD
Swati Sathe, MD, MS
Thomas Seyfried, PhD
Barbara Shapiro, MD, PhD
Evan Snyder, MD, PhD
Cynthia Tifft, MD, PhD, FACMG
Steven Walkley, DVM, PhD
Michael Watson, PhD, FACMG

NTSAD Corporate Advisory Council

Marion Howard, MD, PhD (Chair)
Oved Amitay
Ritu Baral
Michael Gladstone
Kate Haviland
Joan Keutzer, PhD
Greg Licholai, MD

Hope Knows No Boundaries!

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.

national tay-sachs & allied diseases association
susan kahn, executive director (**skahn@ntsad.org**)
joan lawrence, development director (**joan@ntsad.org**)
diana pangonis, family services director (**diana@ntsad.org**)
ingrid miller, office manager (**ingrid@ntsad.org**)
becky benson, conference coordinator (**becky@ntsad.org**)
alison perkus, development associate (**alison@ntsad.org**)

2001 beacon street
suite 204
boston, ma 02135
(617) 277-4463

www.NTSAD.org