

Impact

**National Tay-Sachs
& Allied Diseases Association
of Delaware Valley**

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Jenkintown, PA 19046

www.tay-sachs.org
info@tay-sachs.org

**"TAY-SACHS
AND CANAVAN
TESTING IN EVERY
GENERATION...
FOR GENERATIONS
TO COME."**

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Impact

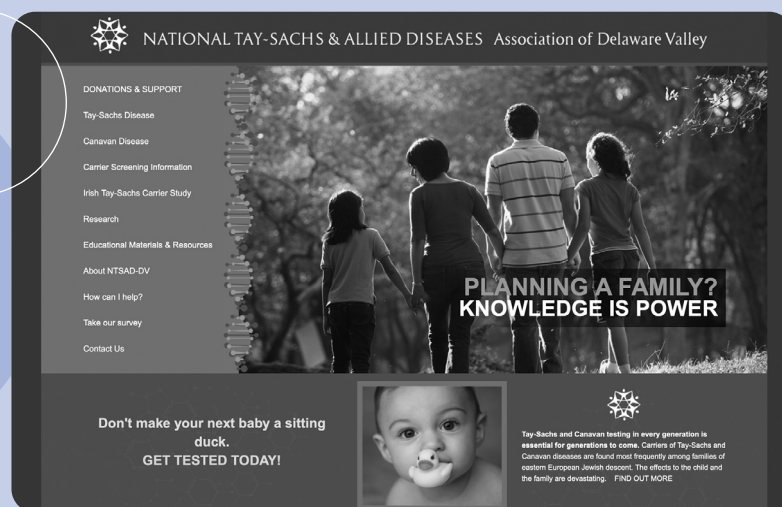
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Impact is a
publication of
National Tay-Sachs
& Allied Diseases
Association of
Delaware Valley

**WINTER
2025**

NEW WEBSITE *COMING SOON*

Another tangible product of your support is our brand-new website currently under development. We expect the new site to launch this spring, and this site would not have been possible without your donations. The website address will remain the same www.tay-sachs.org but will have a refreshed look and updated usability.



THANK YOU FOR YOUR GENEROSITY in returning the enclosed envelope.
You can also make a secure donation online at <http://www.tay-sachs.org/donations.php>

YOUNG LEADERSHIP COMMITTEE

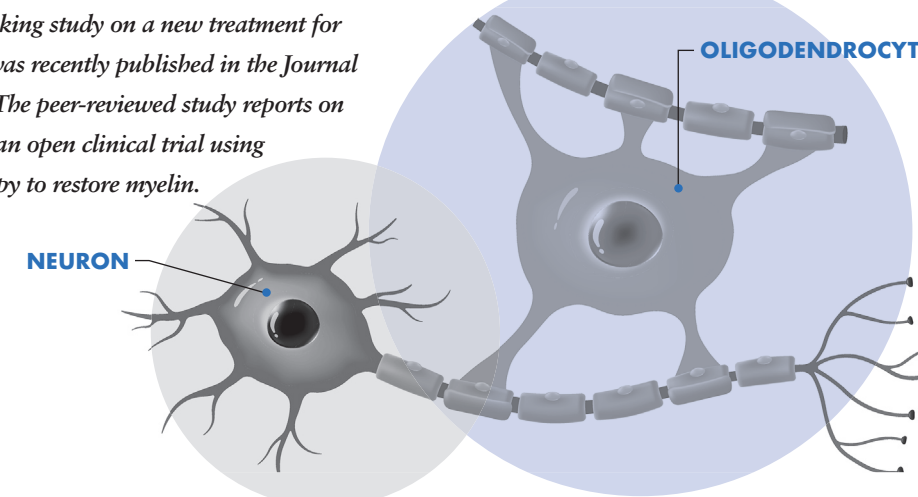
Our Young Leadership Committee continues to engage with the next generation of NTSAD-DV supporters. The YLC is always looking for new members who want to be involved and help us continue our mission.

If you're interested please email
ylc@tay-sachs.org

NATIONAL TAY-SACHS & ALLIED DISEASES ASSOCIATION OF DELAWARE VALLEY (NTSAD-DV)

EXCITING RESEARCH BREAKTHROUGH

A truly groundbreaking study on a new treatment for Canavan Disease was recently published in the Journal Nature Medicine¹. The peer-reviewed study reports on the early results of an open clinical trial using targeted gene therapy to restore myelin.



Like Tay-Sachs Disease, Canavan is an autosomal recessive disease. It is caused when a person inherits both copies of the ASPA gene in each cell that has mutations. The ASPA gene provides instructions for producing an enzyme called aspartoacylase, which is essential for breaking down N-acetylaspartic acid (NAA) in the brain. When the ASPA enzyme is deficient or nonfunctional due to genetic mutations, NAA cannot be properly metabolized. This leads to its accumulation in the brain, which disrupts the formation and maintenance of myelin, the protective sheath around nerve fibers. The progressive destruction of myelin sheaths impairs the ability of nerve cells to send and receive signals effectively. This results in the neurological symptoms characteristic of Canavan disease, such as developmental delays, muscle tone abnormalities, and an enlarged head.

A new clinical trial sponsored by Myrtelle Inc. being led locally by Rowan-Virtua School of Osteopathic Medicine researcher Paola Leone, Ph.D. is using targeted gene therapy to restore myelin, the brain's protective coating, in children with Canavan disease.

Early results show improved brain development and reduced toxic buildup. This new gene therapy is the first designed to target the oligodendrocytes, specialized glial cells in the central nervous system that produce the myelin sheath.

The researchers believe that if they can produce the reconstitution of ASPA function in oligodendrocytes of the brains of Canavan patients, it will allow for NAA metabolism and support myelination/remyelination by the white matter producing cells. Researchers believe this approach offers the safest and most direct therapy for affected individuals.

If their research continues to show such promising results, the potential reaches far beyond Canavan disease. Oligodendrocytes are vital components of the central nervous system, essential for myelination and the efficient functioning of the nervous system, and play a role in other neurological disorders, such as multiple sclerosis and Alzheimer's disease, where myelin damage or loss can lead to impaired nerve function. This new therapy could lead to possible treatments for these and other neurological disorders.

If you would like to keep track of this study and learn more, please visit clinicaltrials.gov and enter the information on the right.

rAAV-Olig001-ASPA Gene Therapy for Treatment of Children With Typical Canavan Disease (CAN-GT)

ClinicalTrials.gov ID [NCT04833907](https://clinicaltrials.gov/ct2/show/study/NCT04833907)

Sponsor Myrtelle Inc.

Information provided by Myrtelle Inc. (Responsible Party)

Last Update Posted 2025-11-14

¹Leone, P., Lober, R.M., Francis, J. et al. Oligodendrocyte-targeted adeno-associated virus gene therapy for Canavan disease in children: a phase 1/2 trial. *Nat Med* (2025). <https://doi.org/10.1038/s41591-025-03919-w>

While the new gene therapy for Canavan is incredibly promising,

NTSAD-DV

remains committed to prevention through screening and education.

YOUR SUPPORT IN ACTION

For almost two years, we have funded through your generous donations a screening fund with our partner, jscreen to access their ReproGen kit with no out-of-pocket cost when used with insurance. For our members who are uninsured or underinsured, we cover the full cost of the kit. The jscreen ReproGen test is easy to use and reliable. The panel is appropriate for pan-ethnic testing. American College of Medical Genetics (ACMG) supports extended carrier screening in all populations.

In last year's IMPACT, we updated you on jscreen's transition to a new structure including a partnership with a commercial testinglab and the creation of an independent 501 c-3. It was a year of significant change and growth for jscreen. We are happy to inform you that this November we were able to fulfill second part of our partnership a community screening event designed to reach individuals at risk pre-conception. With your continued support, we are planning to support additional community screenings and outreach events.

If you are considering getting screened, are a carrier or carrier couple, we urge you to meet with a genetic counselor. Genetic counselors are an important part of the healthcare team. They work with patients and their families to help them understand genetic testing, guide them through the process, and help them make informed choices based on their individual genetic testing results. There are several questions a genetic counselor may help you answer such as:

- What treatments are available for this genetic disease?
- What impact would the genetic test results have on your family?
- What happens if the results are uncertain or inconclusive?
- Should I share these results with family members?

Genetic counselors can go over all the options with you to assist with making the choice that is right for your family. ❄️

NTSAD-DV

directly benefits from your **United Way** donation when you fill out a Donor Choice form with our five-digit code: **00983**. Email info@tay-sachs.org for more information.



NATIONAL TAY-SACHS & ALLIED DISEASES ASSOCIATION OF DELAWARE VALLEY (NTSAD-DV)

EXECUTIVE COMMITTEE MESSAGE

Dear Friends:

Wow. It is truly amazing to think this new clinical trial could reverse the devastating effects of Canavan Disease. As parents of affected children, it has always been our goal to ensure that other families do not have to endure losing a child to one of these diseases. This goal has fueled our efforts for decades. There is a long way to go before this therapy becomes a sustained reality but the promise it holds is truly remarkable.

Every year, we use this message to ask for your continued support and this year is no different. Our plans for 2026 include a continued focus on screening and prevention, but for the first time in the history of our organization we are planning to support research. With every new administration, funding priorities often shift and that has been especially true in the new administration. As parents and as an organization, we feel compelled to do what we can to support the groundbreaking work in this new gene therapy trial. We hope you will join us. Your support makes our work possible in this generation and for generations to come.

Thank you for your generosity.

NANCY DONEGAN MICHAEL J. OSSIP LAURI SUSSMAN SIEGEL



KNOWLEDGE IS POWER – VIRTUAL AVAILABLE

Do you teach a high school biology or genetics course?
Do you work with a community group that could benefit by learning about Tay-Sachs and Canavan diseases and how to get screened?

If so, NTSAD-DV will provide you with a free copy of our educational video "Knowledge is Power – Check Out Your Gene Pool". Our Speakers Bureau for interested schools or organizations is available virtually for the duration of the pandemic. Speakers will present the video to your group, answer questions and provide information about Tay-Sachs and Canavan.

If you would like us to virtually visit your community event or school, or would like more information about our DVD and brochures, please visit our website at www.tay-sachs.org or email info@tay-sachs.org. ❄️

WAYS TO GIVE

There are several options to support NTSAD-DV.
We are grateful for your support.

You can make a secure donation online at <http://www.tay-sachs.org/donations.php>

Return the enclosed envelope

Designate NTSAD-DV as the recipient of your United Way donation Donor Choice 00983



Free Genetic Testing for the first 50 people to register



TIME TO UNZIP THOSE GENES

Reproductive Carrier Screening:
Learn your risk for Tay-Sachs Disease, cystic fibrosis and over 260 other conditions you could pass on to your future children.

How much will it cost? \$0
Thanks to the support of **NTSAD-DV** the full cost of the screening is covered.



National Tay-Sachs & Allied Diseases Association of Delaware Valley
www.tay-sachs.org



1. Scan the QR code to register at jscreen.org
2. Enter to event code: **2025UPENN**
3. Select "Self-Pay" – it's 100% covered, and you won't be charged.
4. Pick up your saliva kit on 11/19/25 at **Penn Hillel**.



215 South 39th Street
Philadelphia, PA

November 19th
11:00am – 1pm and
5:00pm – 7:00pm