NTSAD Community News

Research, Collaboration, and Community

Supporting families is the center of everything we do...

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Advocacy Opportunity for the GM2 Community: First-Ever GM2 PFDD Meeting

Don't miss the opportunity to have your voice heard as we, the GM2 community, address the FDA. **Join us for the first-ever, Externally-Led Patient-Focused Drug Development (EL-PFDD) Meeting with the U.S. Food and Drug Administration (FDA) on February 15, 2024.** The live, virtual meeting is aimed at informing members of the FDA and raising their awareness about GM2 (Tay-Sachs and Sandhoff diseases).

This important meeting, hosted by NTSAD, is free to attend and open to everyone — individuals and families affected by GM2, those who have lost loved ones to GM2, clinicians, researchers, investors, industry partners, and rare allies. Registration is required.

Register Now

The EL-PFDD Meeting will feature moderated conversations and clinical overviews of the three onset levels of GM2, statements from individuals and family members who will speak about the impact of GM2 on their lives, their experiences with disease management, and their hopes for accelerated drug development.

"The patient perspective is critical in helping FDA understand the context in which regulatory decisions are made for new drugs. PFDD meetings give FDA and other key stakeholders, including medical product developers, health care providers, federal partners, ***an important opportunity to hear directly from patients, their families, caregivers, and patient advocates about the symptoms that matter most to them, the impact the disease has on patients' daily lives, and patients' experiences with currently available treatments.*** *This input can inform FDA's decisions and oversight both during drug development and during our review of a marketing application."* – FDA

Learn more about the first-ever EL-PFDD meeting for GM2 at our community webinar on Wednesday, January 10, 2024, 12-1 pm Eastern. **Register now!**

FDA Establishes Advisory Committee for Genetic Metabolic Disease Treatments

The US Food and Drug Administration (FDA) has announced the establishment of a new advisory committee to evaluate potential treatments for genetic metabolic diseases. The Genetic Metabolic Diseases Advisory Committee will advise the FDA on products used for the diagnosis, prevention and treatment of genetic metabolic diseases under the Division of Rare Diseases and Medical Genetics.

Described as *"an essential part of the FDA's work, advisory committees allow the FDA to receive invaluable input from clinicians, industry experts, academia, patients, caregivers and other external stakeholders when evaluating the potential benefits and risks of a new therapy,"* said Patrizia Cavazzoni, Director of the CDER.

The new advisory committee will help the FDA increase their understanding of rare diseases, and we hope to accelerate approvals for potential treatments for Tay-Sachs, Canavan, GM1, and Sandhoff diseases.

Learn more.

Azafaros Study for GM2: Now Fully Enrolled

AZAFAROS B.V.'s Phase 2 RAINBOW study, evaluating nizubaglustat in Tay-Sachs, Sandhoff, and Neimann-Pick-C patients, is now fully enrolled.

Azafaros' Chief Executive Officer Stefano Portolano, M.D., said *"Exceeding the recruitment goal is an important step forward in our ambition to provide a new therapeutic solution for these rare lysosomal storage disorders, where patients have no or limited treatment options. Our team has worked tirelessly to reach this point and the focus is now on preparing the next steps for two Phase 3 studies. We would like to thank all patients, their families, patient organizations and clinicians for their participation and support in this important research."*

Thank you, Azafaros, for your continued commitment to finding effective treatments for rare diseases!

Read the press release.

Finding A Community -- By Lee and Lori Greenwood

"When we got Noa's diagnosis, we were wholly unprepared to be told that our daughter might not live to see her 10th birthday."

Getting this kind of news is devastating. *"The few resources available online offer little to a newly diagnosed family.* ***But then we found NTSAD. We found a community — our community.*** *We were immediately connected to other families who knew exactly what we were feeling: grief, shock, desperation, and deep love for our children. The NTSAD community quickly became more like a family, which was exactly what our family needed."*

Your support makes a difference in the lives of rare individuals and families.

"We found resources — ones to support our older daughter Max, a super sibling, to support us as parents, and to help us better understand our daughter's disease and how to care for her complex needs. ***We found an organization that funds medical advancements and the vital research that's gotten us and Noa where we are today."***

You can help more children like Noa by making a gift to support family services and research.

Donate Now

Grief and The Holidays

NTSAD knows that the holiday season can be extremely difficult for members of our community. If you are experiencing grief, please be kind to yourself. This season is all about love, and grief is an expression of love preserving through loss. It's okay to not be okay.

NTSAD's Rare is Real Sweatshirts

Get ready for the New Year and Rare Awareness Day in February by sporting a NTSAD zip-up sweatshirt that affirms Rare is Real! You can raise rare awareness while also supporting rare families. A portion of the proceeds go to programs for families and research.

Purchase a Rare is Real Sweatshirt!

Why I Give to NTSAD (and from my IRA) -- By Judy Kaplan

After several years of trying to find a diagnosis, our precious 17-year-old daughter, Linda, was finally diagnosed with Late

Onset Tay-Sachs (LOTS) in 1988. We were heartbroken, but relieved to have a diagnosis.

In the days before the internet, it was exceedingly difficult to find any information, especially for a condition that had only been recognized a few years earlier. Fortunately, a knowledgeable genetic counselor told us about NTSAD.

At the time, there were seven other people with LOTS connected with NTSAD, and my family contacted all of them. There was little medical knowledge about treatments available at the time, so families shared what seemed to work, or not work, based solely on their experience.

We attended our first NTSAD Annual Family Conference in 1989, and many more since then. We realized that it was challenging for some NTSAD families to find money to travel across the country and stay in a hotel, all while caring for sick and disabled children and adults. So, we started the annual Helping Hands raffle, with all the proceeds being used to provide assistance for families who wanted to attend the conference.

With the help of NTSAD, our little group reached out to the practitioners and researchers to get them interested in this ultra-rare condition. We quickly discovered that research required funding, and we needed to help raise funds.

All of us donated money then, and I continue to give every year in memory of my beloved Linda to support other families like mine and to advance research to find treatments for this horrible disease. Now that I am retired, I give through my IRA (Individual Retirement Account) to maximize both my donation and my tax benefits.

As you may know, since the 2017 change in U.S. federal tax policy, you can no longer include charitable donations -- together with medical expenses and several other categories -- as itemized deductions, unless the total exceeds the amount of the standard deduction. To me, this seems unfair to charitable organizations like NTSAD that rely on donations to stay afloat, as some people prefer to receive a tax benefit and may give less via post-tax income.

But if like me, you are living in the U.S., are retired, and have an IRA, you can take a charitable deduction on your federal taxes when you donate via your IRA.

Here is how to use pre-tax dollars for charitable donations:

1. Contact the financial institution that holds your IRA funds and request a QCD (Qualified Charitable Distribution) Form.
2. Enter your personal information, the charitable organization's information, and the amount of money you wish to give, then return the form to your financial institution. (Note: the payment must go directly to the addressee or the charitable organization to qualify; do NOT have the check sent to you, or any other person.)
3. Make sure you keep a copy of the completed QCD form with your tax information for the current year.
4. At the end of the year, include the QCD with the rest of your tax information, and make a point of notifying your tax preparer about this donation.

Now you have two choices-- feel good about your donation and the money you saved, or feel even better and increase your donation to include the amount of your tax savings! That's what I do. Join me and make a donation using your IRA. **Together, we can make a difference for families and advance research.**

NTSAD leads the worldwide fight to treat and cure Tay-Sachs, Canavan, GM1, and Sandhoff diseases by driving research, forging collaboration, and fostering community. Supporting families is the center of everything we do.

Donate

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