NTSAD Community News

Research, Collaboration, and Community

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

April

April 2024

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NTSAD Community Comes Together

More than 300 people, including 74 families, gathered for the 46th Annual Family Conference in Chicago from April 11 to 14. Rare children, adults, and their families travelled from across the United States from Alaska to North Carolina and internationally from Argentina, Canada, Chile, Denmark, Israel, Mexico, and Slovakia to attend the four-day event, along with clinicians, researchers, and members of industry.

As one attendee shared, ***"You all prepared this conference with the magic ingredient: Love! And it was noticeable in the atmosphere of the conference. I feel blessed I could attend."***

On Thursday, rare adults and families participated in support sessions, including 17 people in attendance for the first time, and five families who primarily speak Spanish. A huge thank you to interpreter Anabella Roman, who generously volunteered her time and expertise.

On Friday's Research Day, NTSAD CEO Kathy Flynn and Research Director Valerie Greger, PhD, gave an overview of NTSAD's research priorities and recent investments. Then, former NTSAD Executive Director Sue Kahn moderated the "Ask the Industry Expert" panel who discussed the current climate in research and drug development for Tay-Sachs, Canavan, GM1, and Sandhoff diseases. Panelists included: David Rintell, EdD, of Aspa Therapeutics; Gisela Linthorst of Azafaros; Taylor Fields of IntraBio; Mathias Schmidt, PD, PhD, of JCR USA; Olga Flamini, MD, PhD of Myrtelle; and Cristina Cardoso of Sanofi.

On Friday afternoon our community gathered to support more than 50 bereaved individuals and families as they honored their loved ones. We said their names, lit candles, and created beautiful bouquets of tulips, which symbolize perfect love. Thanks to rare moms Monica Gettleman, Bonnie Davis, Amber Franzen, and Becky Benson for planning the thoughtful tribute.

On Friday, 15 members of the Late Onset group enjoyed a bus tour into Chicago and stopped at Navy Pier. Throughout the weekend, loving professional care was provided to 26 rare children by incredible Camp Snuggle and Camp Active volunteers. Special thanks to Holly Stringer, Caitrin Alexander, and Karen Horton for making these camp experiences possible as well as fun for our medically fragile, little ones and giving parents respite so they could attend sessions.

Our youth group, consisting of 15 super siblings ages 11-17, enjoyed field trips to Wrigley Field, the Bean, Navy Pier, the Shedd Aquarium, and the Field Museum over the weekend. Thanks to our volunteer chaperones Loren Benson, Craig Franzen, and Rod Marquardt. Our younger group of 20 super siblings also had a blast at Camp Sunshine, organized by KiddieCorp. Special thanks to Rare Dad Rudy Suarez for securing personalized surprises for all the kids.

On Saturday evening, Crystal Villalobos and Dan Greenberg were recognized for their efforts to spread awareness, advocate for rare, and advance research. Photographers Sarah Mattingly and Jamie Carpenter (from Living in the Light) gave 43 families and adults professional portraits. Local firefighters took photos with the children, and Gene the therapy dog also made a return visit. We ended the day cheering for all the children as they paraded through the hotel leading everyone into dinner.

Thank you to all who attended and supported the Conference. We greatly appreciate our 2024 sponsors and donors, who made the conference experience possible, including providing 51 Helping Hand grants to families.

Thank you to our 2024 Sponsors!

Presenting

Anonymous

THE DOYLE FOUNDATION

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Lauren Celano

Stanley Cohen

Bonnie and Barry Davis
in Loving Memory of Adam Davis

Monica and Gary Gettleman
in Memory of Brooke Chase Gettleman

Jaxson's Train of Hope — Connect for a Cure — Derail Rare Diseases

Staci Kallish, DO

NTSAD Delaware Valley

Susan and Alan Roden and Family

47th Annual Family Conference: Dallas, Texas, 2025

Save the date!! **NTSAD's 47th Annual Family Conference will take place on April 24-27, 2025, in Dallas, Texas!** Yee-haw!

We need your help! Send your "Dallas" theme suggestions for the conference to Family Services Manager Becky Benson at **becky@ntsad.org**.

NTSAD Invests in GM2 Research in 2024

NTSAD's Research Initiative along with the Cure Tay-Sachs Foundation has awarded three researchers, Angela Gritti, PhD, Amanda Gross, PhD, and Jessica Larsen, PhD with a total of $100,000 in funding for two research projects to advance treatments for GM2 (Tay-Sachs and Sandhoff diseases).

Angela Gritti, PhD
IRCCS Ospedale San Raffaele (OSR), San Raffaele
San Raffaele Telethon Institute for Gene Therapy (SRTiget)

Enhancing the Therapeutic Potential of Hematopoietic Stem Cell Gene Therapy to Treat GM2 Gangliosidoses

Dr. Gritti's proposal is to improve hematopoietic stem cell gene therapy for GM2 gangliosidosis. This kind of treatment using blood cell production has shown tremendous promise in other lysosomal storage disorders. **Strong preclinical data from this study could provide additional treatment options for Tay-Sachs and Sandhoff diseases in a relatively short time frame.**

Amanda Gross, PhD
Auburn University College of Veterinary Medicine

Jessica Larsen, PhD
Clemson University College of Engineering

Nanoparticle Distributed Intravenous Enzyme Replacement Therapy (NanoDIVERT) for Tay-Sachs and Sandhoff Diseases

A drawback of traditional enzyme replacement therapy (ERT) for neurodegenerative disorders such as GM2 gangliosidosis is the blood brain barrier prevents the enzyme from reaching the brain where it is needed most. Drs. Gross and Larsen's proposal explores the use of nanoparticles to transport enzymes through the blood brain barrier. **If successful, this research may open the door to developing an alternative, non-invasive approach for the treatment of Tay-Sachs and Sandhoff diseases.**

In 2022, Amanda Gross, PhD, received a grant from NTSAD for her work in developing gene therapy for the treatment of feline GM1. Dr. Gross recently shared positive outcomes of her work at NTSAD's 46th Annual Family Conference.

*Special thanks to the **Cure Tay-Sachs Foundation** for co-funding these projects and continued partnership.*

2024 Imagine & Believe: Honoring the Buryk Family

Each year at NTSAD's signature fundraising event, ***Imagine & Believe,*** we recognize an individual or group who makes a significant and positive impact on the rare disease community. **We proudly announce this year's honorees are Buryk family members–parents Alexis and Bill, and twin daughters Katie and Allie, who both have Late-Onset Tay-Sachs disease, and eldest daughter Elizabeth and her husband Jonathan Rego.**

In 2014, the Buryk family launched the Katie & Allie Buryk Research Fund at NTSAD. To date, the Buryk Research Fund has raised nearly $1,100,000 for research for Late Onset Tay-Sachs and Sandhoff (LOTSS) diseases with the help of more than 2,400 donors. You may have seen the Buryk Family's powerful ad which runs quarterly in *The New York Times.*

When launching the Katie & Allie Buryk Research Fund a decade ago, Katie shared what motivates her, ***"I have decided to go public and try to raise money, awareness and mount a search for a cure. Sure, it's for me and Allie, but it is also for infants and kids who aren't as lucky as we are. And it's for the families who must embark on this difficult, sad, and baffling journey."***

The Buryk Research Fund has supported research grants, helped NTSAD hire our first Research Director, and hosted six Late Onset Think Tanks meetings to foster collaboration and innovation in advancing research and clinical trials. The Buryk Family has made a substantial impact on the rare community and helped to drive not only research for Tay-Sachs and Sandhoff diseases, but awareness for all onset levels of Tay-Sachs and Sandhoff as well as Canavan and GM1 diseases. The family's *New York Times* ad has helped at least two women in our community receive a proper diagnosis.

Join us as we honor the Buryk Family and Save the Date for *Imagine & Believe* on November 7, 2024, at Le Meridien Hotel in Cambridge, Massachusetts.

Fifty-Seven Fridays by Rare Mom Myra Sacks

Rare Mom Myra Sacks recently released a memoir about the short, but well lived life of her daughter, Havi.

"Life is unfolding as planned for Myra Sack and her husband Matt until their beautiful year-old daughter Havi is diagnosed with Tay-Sachs disease and given only a year to live. Myra and Matt decide to celebrate Havi's short life and vow to show her as much of the world as they can, surrounded by friends and family who relocate to be in Havi's orbit. Tapping their Judaism, they transform Friday night Shabbats into birthday parties—"Shabbirthdays"—to replace the birthdays Havi will never have."

Read this beautiful and heartbreaking story of life and love.

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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