



# NTSAD Community News

Research, Collaboration, and Community



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

# July

July 2022

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Dear Friends,

We hope you are enjoying the summer. How quickly it is passing by! Earlier this month, we were thrilled to connect in-person with families, researchers, clinicians, and industry members at NTSAD's 44th Annual Family Conference in Denver. It was extraordinary to welcome 175 people, many attending the four-day event for the first time, including myself!



I will never forget the pure joy that danced across the faces of families as they reunited after being apart for three years – and the warm welcome new families received from the NTSAD Community. There was an electricity in the air from the moment everyone arrived until the last car drove away.

Thank you to everyone who made the trip to Denver and to all those who supported this year's conference.

**To all our families and friends who couldn't attend this year's Conference, we hope to see you next year for the 45th Annual Family Conference, which will take place June 1-4, 2023, in Reston, Virginia!**

After returning from Denver, I represented NTSAD at the Chief Patient Officer Summit in Boston and participated in a panel about how to building long-term relationships between patient advocacy organizations and industry. We discussed the importance of transparent communications with patients and families, the tenets of a good relationship (input from families!), and best practices benefit patients and families.

I feel fortunate anytime I can speak about NTSAD's rich history of supporting families and forging collaboration with researchers, industry, and other stakeholders to advance our mission. We thank you for being a part of our Rare Community and joining us as we strive toward effective treatments.

Sincerely,

Free Carrier Screening

Auction Items Needed!



Kathleen M. Flynn  
Chief Executive Officer

## Together Again at the 44th Annual Family Conference



Thanks to the families, speakers, attendees, and sponsors for making NTSAD's first in-person conference in three years possible and so very special. The 44th Annual Family Conference in Denver, Colorado brought our Community together again, following two years of virtual conferences, and our families are particularly grateful. For many of our newly diagnosed families, the conference was the first time they could talk face-to-face with other families who truly understand.

Mariam, mom to Alya, shared her appreciation for the connections she and her family made while attending their first conference.

***“We had such a memorable time. I feel blessed to have met all these wonderful families and friends. Thank you so much for everything you and your team did for our family. We couldn't have asked for anything more! We would not have been able to meet if it wasn't for a Helping Hand grant! I feel so rejuvenated and ready for what the next year brings us.”***

For four days, families participated in health management sessions, peer support groups, and research updates, as well as enjoying each other's company during meals and in the family lounge. Our Late Onset community came together to support one another and share their stories and experiences. Affected children attended Camp Snuggle and Camp Active, while their healthy siblings attended their own camps and retreats. Affected children were lovingly held and made art projects and keepsakes to take home with the help of volunteers, many of whom honor lost loved ones by caring for the children attending camps. **For many parents, camps are the first time**



**they have been able to leave their child for a few hours, receive support themselves, or even take a break, while their children laugh and play and make friends with kids just like them.** Alya formed an instant friendship with Maddie and Lily, and the three were inseparable throughout the week. Maddie and Lily are pictured below playing in Camp Active.



There are camps for healthy siblings, too. Younger children attended Camp Sunshine playing games and having fun, and older siblings visited The Denver Museum of Nature and Science, the Denver Zoo, and Meow Wolf.

On Friday, families joined clinicians, researchers, and industry members to hear updates on current clinical trials and upcoming research projects. Thank you to all the speakers and facilitators! Stay tuned for research updates from the Conference.

On Saturday, a moving Commemoration ceremony was held for bereaved families who remembered and honored loved ones gone too soon by lighting a candle when their loved one's name was announced. This year, families placed a flower representing their loved in one unified, memorial bouquet. Families were also given forget-me-not seeds to plant when they returned home.

**The Conference ended on a high note singing “Happy Birthday” to Alya, who celebrated her 9th birthday surrounded by new friends.** (See Alya looking regal in her pink party dress next to Mom Mariam.)



Check out more #AFC2022 photos on NTSAD's [Facebook](#).

## Imagine & Believe 2022

Join NTSAD for a night of *Imagining & Believing* in a world free from rare diseases. **On Thursday, November 10, 2022, the NTSAD Community will gather at the Royal Sonesta in Boston for the first in-**

**person *Imagine & Believe* event in three years to commemorate NTSAD's 65th Anniversary.** At the event, which raises funds for programs and services, NTSAD will honor research pioneer Guangping Gao, PhD, for revitalizing gene therapy and seeking potential treatments for rare diseases, including his research toward a gene therapy for Canavan Disease.



*“I hope you will join me and the entire NTSAD Community as we commemorate 65 years of supporting families, providing education, raising awareness, and driving research. Together, we hope for a future with effective treatments and cures for Tay-Sachs, Canavan, GM1, and Sandhoff diseases.”* -Oralea Marquardt, LCSW, NTSAD Board President

[Learn more about Imagine & Believe sponsorship opportunities here.](#)

[Watch last year's Imagine & Believe videos.](#)

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## NTSAD Welcomes New Board President Oralea Marquardt



Oralea and Rod Marquardt's lives changed forever when in 2007 their second child, William, was diagnosed with GM1 gangliosidosis. Feeling alone and overwhelmed by emotions and medical decisions, they found their way to NTSAD in search of resources to help them care for their son.

Through NTSAD, Oralea's family found compassion, understanding, and support from other families and the NTSAD family services team. Two years after William passed, Oralea was inspired to join the NTSAD's Board of Directors, so she could support other rare families.

Soon, she was tapped to become NTSAD's next Board President.

Oralea's experience caring for William led to her partnership with improving palliative care in her Florida community, as well as a new career path. Recently, Oralea became a Licensed Certified Social Worker (LCSW), where she works to advocate for families like hers. Fittingly, Oralea led her first Board meeting as President on William's 17th birthday.

*"It is an honor to serve as Board President at such an exciting time for NTSAD. Sixty-five years ago, there was little knowledge about Tay-Sachs, Canavan, GM1, and Sandhoff diseases, let alone potential treatments. Now, there are clinical trials underway for each of these diseases. Although the NTSAD Community is in this new era of clinical trials, there's still so much more work to do. NTSAD remains committed to finding effective treatments and a cure for families."* -Oralea Marquardt, LCSW, Board President

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## Thank You for Your Service

Every June, NTSAD reviews and renews its Board leadership, acknowledging departing members for their service, and at the same time welcoming the next generation of leaders. **NTSAD is incredibly grateful to outgoing Board President Staci Kallish, DO.** Staci has been involved with NTSAD since she was a young girl along with her mom, Meryl Kallish, a former Board President, herself. Inspired by NTSAD families from an early age, Staci became a geneticist.

At the 2022 Family Conference, new NTSAD Board President Oralea Marquardt thanked Staci for leading NTSAD through the COVID-19 pandemic and the start of clinical trials. Staci will continue to serve on the Board and its Executive Committee as Immediate Past President. Thank you, Staci!



On behalf of NTSAD families, NTSAD thanks **Karen Grinzaid, MS, CGC, CCRC**, Executive Director of JScreen, (a genetic screening and education company) who will continue to contribute to the Research Committee; **Sophia Pesotchinsky**, who remains on the Research Committee and also is Mom to Vera who has Late Onset Tay-Sachs disease; and **Marion Yanovsky**, a long-time volunteer, who serves as chair to the New York Area Fund and continues committee work for NTSAD. **We thank Karen, Sophia, and Marion for their many years of service on the Board of Directors and**

dedication to families. NTSAD is a better organization and a stronger Community thanks to them!

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## Welcome New Board Members

Please join us in welcoming new NTSAD Board members Dawn Laney, Jon Lawrence, Susan Roden, and Deborah Sanders.

**Dawn Jacob Laney, MS, CGC, CCRC**, is a genetic counselor, instructor, researcher, director of the Genetic Clinical Trials Center, and program leader of the Lysosomal Storage Disease Center at Emory University, Department of Human Genetics. Dawn earned her master's degree in medical genetics from the Sarah Lawrence College. In 2002, she received her certification from the American Board of Genetic Counseling and joined the Emory where she has been involved in follow-up of abnormal metabolic newborn screening for the state of Georgia, development and management of a genetic infusion center, and lysosomal storage disease clinical care and research.

**Jon Lawrence** joins the board in honor of his son, Cameron, who has Canavan disease, and brings his legal expertise to the Board. Jon runs his own practice and provides consulting services to other attorneys and law firms, including development of litigation and trial strategy. Jon received his JD from Florida State University College of Law Tallahassee.

**Susan Roden**, a retired IBM executive, has been involved with the NTSAD New York Area Chapter since childhood and is a second-generation volunteer. Susan has supported many fundraising efforts including walks, golf outings, annual calendar, cocktail receptions, and special events held in New York. She also has focused on educational initiatives to spread awareness of carrier screening. Currently, Susan serves as a member of the NTSAD NY Area Fund Committee.

**Deborah Sanders** is the Vice President, Global Customer Care and Customer Ambassador at Avid, a software development company. She has a depth of professional experience in leading large, multi-functional, multi-cultural customer support and delivery teams during organizational growth and change. She has worked in an array of industries including SaaS/Computer Software, Access Networking, IT, Industrial Automation, Life Safety, Regulatory Compliance, and Hydrocarbon Chemicals Manufacturing.

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## Heather Gray-Edwards among Five Researchers to Join Advisory Council



Nearly eight years ago, NTSAD created a **Scientific Advisory Council (SAC)** bringing together pioneering researchers who committed their careers to advancing clinical research and gene therapy to advise NTSAD's research strategy and investments. To ensure the current research focus of clinical development, early diagnosis, newborn screening, and translational research is reflected in the expertise of the SAC membership, current Co-chair **Frances Platt, PhD**, University of Oxford, along with NTSAD CEO Kathy Flynn and Research Director Valerie Greger, PhD, invited **Heather Gray-Edwards, DVM, PhD, UMass Chan Medical School to join as SAC Co-chair.**

**Four additional members also joined the SAC: Doug Martin, PhD**, Auburn University; **David Pleasure, MD**, UC Davis Health; **Miguel Sena-Esteves, PhD**, UMass Chan Medical School; and **Christopher Stephen, MBCHB**, Mass General Research Institute.

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## Risa Asnen Goes Above and Beyond

Each year, NTSAD recognizes an individual who is not directly affected by a rare disease, and yet has dedicated countless hours of service and support to our Community. **This year, we acknowledge Risa Asnen for her extraordinary service with the Above and Beyond Award.**



For more than 40 years, Risa Asnen has been part of the NTSAD Community serving as a volunteer and then as a Board member. For decades, she was the Vice President of the Annual Family Conference, creating a place for NTSAD families to receive intensive support and make lasting friendships. Risa managed everything for the event finding accessible locations and hotels, coordinating meals, creating sessions with the Conference Committee, and making sure each family received what they needed. Risa has been a true, life-long volunteer, and a cherished member of the NTSAD Community. We thank and recognize Risa for her years of service and unrelenting dedication to families. Risa has shown what it truly means to go Above and Beyond!

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## NTSAD Power of One Award: Daniel and Patricia Lewi



NTSAD recognized CATS Foundation Founders Daniel and Patricia Lewi with the 2022 Power of One Award. They established the [Cure and Action for Tay-Sachs Foundation \(CATS Foundation\)](#) in June 2011, after their daughter Amelie was diagnosed with Tay-Sachs at 15 months of age. At the time, there was no charity based in the United Kingdom (UK) dedicated to providing support for families affected by Tay-Sachs or Sandhoff diseases.

Dan and Patricia continue to honor their daughter, who passed away in December 2017 at the age of eight, by providing support to families in the UK and beyond. For more than a decade, the CATS Foundation has served as a tremendous resource for families while also raising awareness of Tay-Sachs and Sandhoff diseases and funds for research. NTSAD is proud to honor Dan and Patricia for their service to families and their ongoing collaboration to find effective treatments and a cure.

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## Auburn University's Research Has Positive Effects on GM1 Patient



Jojo, who is 13 years of age, is the first child with GM1 to participate in a clinical trial based on Auburn University's research and has shown signs of improvement.

Jojo's mother shared, ***"Jojo was very ill when she was treated with gene therapy in 2019. She had great trouble swallowing and eating, and she wasn't able to walk. Since treatment, Jojo eats normally and is at a normal weight for her age. She still has some challenges, but she's made great improvements in many ways."***

Researcher Doug Martin, PhD, Director of Auburn's Scott-Ritchey Research Center in the College of Veterinary Medicine and a professor in the Department of Anatomy, Physiology, and Pharmacology, who hoped to stop the disease's progression, confirms, ***"She is showing real signs of improvement."***

The current NIH trial, which includes 11 children, is the first of three trials underway. Dr. Martin and his fellow Auburn researchers are moving forward with additional research as well, hoping to add to the knowledge base for GM1 and the related GM2 diseases (Tay-Sachs and Sandhoff).

[Read the full article.](#)

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## Manning Family Honors Son Dylan and Support NTSAD Families

For nearly two decades, Brian and Sherri Manning have honored their son Dylan James Manning, who had Tay-Sachs disease, by hosting a golf tournament outside Philadelphia. **This year was the 19th Annual Drive FORE Dylan and, with the help of their loyal supporters, the Mannings raised nearly \$40,000 for NTSAD family programs and services.** To further honor Dylan's legacy, the Manning Family works to spread kindness all year long through a kindness campaign. This past spring, they made a personal connection with the teachers and students of Riverside Elementary School in neighboring New Jersey. Brian and his daughter Mia traveled to the school for a special day to support the students' efforts to spread kindness in their community and to share Dylan's story. Thank you to the Manning Family, for their extraordinary efforts to raise awareness, funds, and spread kindness. (Brian and Sherri pictured below with daughter Mia and sons Brady and Devin.)



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## Day of Hope 2022

Each year NTSAD families, friends, and rare allies come together for a "Day of Hope" and host events to raise awareness and critical funds for research leading to effective therapies for our rare diseases. This year's Day of Hope is September 17, 2022. Starting in the summer and running through the fall, families are hosting walks, runs, dance parties, pancake breakfasts, trivia nights, golf tournaments, Facebook fundraisers, t-shirt campaigns, and more. It's a great way to engage your Community to support you, your child, and every NTSAD family. Fun for employees and industry partners, too!



The NTSAD Staff are here to help you! We can provide ideas, guidance, and giveaways for attendees. It can be as easy as launching a Facebook fundraiser, or creating your own "TEAM" Day of Hope t-shirt that you design with NTSAD's partner vendor, Bonfire. This fundraising option will be available in August! All you do is design a shirt, then share the link and friends order and pay online.

Bonfire ships the shirts directly to your family and friends and proceeds go to NTSAD and Day of Hope.

Since the first Day of Hope held in September 2011, more than \$560,000 has been raised for NTSAD Research Initiatives leading to potential new therapies for Tay-Sachs, Canavan, GM1, and Sandhoff diseases.

**[Interested in learning how to get involved this Day of Hope? Register now for an informational zoom meeting on Tuesday 8/9 at 6:00pm EDT!](#)**

**[Support Day of Hope 2022 and Make a Gift Now.](#)**

**[Purchase a Day of Hope t-shirt!](#)**

**[Guide to Planning a Day of Hope Event.](#)**

For assistance on hosting a family event or corporate activity email Development and Communications Manager Sydnie Dimond at [sdimond@ntsad.org](mailto:sdimond@ntsad.org)

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## Thank you to 2022 Annual Family Conference Sponsors

Thank you to the 44th Annual Family Conference Presenting Sponsors, [The Doyle Foundation](#) and [Sanofi](#), and all our sponsors, whose support provide families with connection, community, and support.

***“I'm so incredibly thankful for the family conference and the Helping Hand grant. We would not have been able to make the trip without the help. We owe so much to NTSAD staff and families for all their support this past year.” – Kim Rudness, Mom to Greyson***



### PRESENTING

[The Doyle Foundation](#)

[Sanofi](#)

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Anonymous

Drs. Roselyn and Edwin Kolodny, Rare Humanitarian

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### Connection

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In Memory of Olly Belle  
Chapman

In Memory of Brooke Gettleman

[JScreen](#)

Martha\* and Dr. Ron Kleinman

\*NTSAD Board Member

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## JScreen Offers Free Carrier Screening for Affected Families

For siblings and extended relatives in families affected by rare diseases, fear of the unknown can prevent planning or expanding their families. **NTSAD hopes to empower families by offering free comprehensive carrier screening services to family members of an affected child or individual.** Thanks to the Evan Lee Ungerleider Fund of NTSAD, the NTSAD New York Area Fund, Mathew Forbes Romer Foundation, and the Canavan Foundation for making free screening possible through JScreen and giving families the information they need to safely plan and expand their families.

**[Go to JScreen.org and use code NTSAD22 for a ReproGen test at no cost to you. Insurance information required.](#)**

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## Support NTSAD through a Gift to the *Imagine & Believe* Auction!

NTSAD is seeking donations of items and experiences for the silent auction at *Imagine & Believe* as well as volunteers who can help solicit and secure them. Items such as bottles of wine; gift certificates for dining at restaurants and chef's tables; airline vouchers; travel opportunities including vacation stays and getaways; theater (Broadway!), concert and sporting event tickets and memorabilia; unique experiences; jewelry; and spa packages will help us to raise money for programs and services for families. Event guests live in Boston and out-of-town, and we are seeking physical items that may be easily shipped as well as gift cards and opportunities.

**If you are interested in donating an item or volunteering for NTSAD's auction committee, please contact Development and Communications Manager Sydnie Dimond at [sdimond@ntsad.org](mailto:sdimond@ntsad.org)**

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**NTSAD leads the worldwide fight to treat and cure Tay-Sachs, Canavan, GM1, and Sandhoff diseases by driving research,**

**Donate**

**forging collaboration, and fostering community. Supporting families is the center of everything we do.**

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