NTSAD Community News

Research, Collaboration, and Community

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

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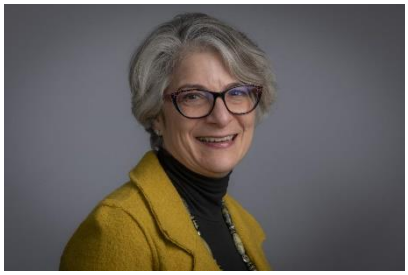
Honoring Patient Advocate Jayne Gershkowitz



Each year at NTSAD's signature fundraising event, *Imagine & Believe,* we recognize an individual who makes a significant and positive impact on the rare disease community. **We proudly announce this year's honoree, Jayne Gershkowitz, Chief Patient Advocate at Amicus Therapeutics, a trailblazer in patient advocacy and a remarkable individual who dedicated her career to championing children, adults, and families affected by rare diseases.**

A long-time patient advocate, Jayne joined Amicus Therapeutics in 2006, where she develops and executes global strategies that ensure people living with rare diseases and their families remain at the core of all of Amicus' operations. Jayne founded the company's Patient Advisory Boards program, which helps give voice to the concerns of patients, families, and caregivers, and she leads Amicus' Public Policy work to advocate for policies that advance access to diagnosis, drug development, and treatment to better satisfy unmet needs among those living with rare diseases.

After many years in corporate marketing communications and nonprofit management, Jayne began her extraordinary career in patient advocacy leading NTSAD as Executive Director from

1998-2006. She broadened programs and services for families and was a driving force in the launch of NTSAD's Research Initiative, creating the path for today's advancements.

A tireless and recognized expert, Jayne co-founded Professional Patient Advocates in Life Sciences (PPALS), an organization that provides authentic patient advocacy education for biotech and pharmaceutical professionals. Throughout her career, Jayne has been honored for leadership efforts, speaks frequently at industry conferences, and serves on several boards.

We hope you join us as we celebrate Jayne on November 9, 2023, at *Imagine & Believe,* held at the Royal Sonesta Boston. Mark your calendar and save the date!

Read more about Jayne and *Imagine & Believe* here.

NTSAD Supports GM2 Trial

One of NTSAD's main priorities is advancing research and clinical development for Tay-Sachs, Canavan, GM1 Gangliosidosis, and Sandhoff diseases. In February, NTSAD led a collaborative funding effort to ensure the continuation of UMass Chan Medical School's GM2 gene therapy trial following the dissolution of the biotech company that was previously running the trial.

Terence R. Flotte, MD, the Executive Deputy Chancellor, Provost & Dean of UMass Chan Medical School, expressed the importance of this unified support.

"We recently have treated the seventh patient in the GM2 (Tay-Sachs and Sandhoff disease) trial and the ninth overall with the GM2 vector. Each of these infants has only a very brief window of time to receive gene therapy, after which time they lose the ability to respond. We are incredibly grateful to the National Tay-Sachs & Allied Diseases Association, Matthew Forbes Romer Foundation, Cure Tay-Sachs Foundation, and Blu Genes Foundation for providing the funding that has enabled this trial to continue. *We remain fully committed to bringing the best available technology to as many infants as we possibly can and to seeing this program through to the end."*

NTSAD is proud to support this trial, and with additional data, help to move forward the development of treatments for Tay-Sachs and Sandhoff diseases. We look forward to hearing an update from Dr. Flotte at the Annual Family Conference.

Positive Data in Canavan Patients Dosed with Myrtelle's Gene Therapy

Myrtelle, a gene therapy company, recently announced encouraging initial data and a favorable safety profile 6-month post-treatment in the eight patients dosed with the Myrtelle's investigational gene therapy for Canavan disease.

The six-month functional and anatomic data observed in these patients is encouraging in showing positive changes in the patients' motor, language, cognitive, and visual skills," explains Chris Janson, MD, Principal Investigator and Assistant Professor of Neurology and Neuroscience at Wright State University Boonshoft School of Medicine, and Director of Human Gene Therapy Center at Wright State Neuroscience Institute in Dayton, Ohio.

Read the full press release.

Lysogene Reorganization Proceedings Continue

Lysogene, a gene therapy company based in France, who was focused on clinical development for gene therapy for GM1 gangliosidosis among other diseases, announced that no takeover offer was filed, and reorganization hearings will continue.

Read the press release here.

45th Annual Family Conference

NTSAD's Annual Family Conference provides affected families and individuals with the rare opportunity to connect with people who truly understand, learn about the latest research and symptom management approaches, and support one another.

Hear about the conference experience directly from families in NTSAD's new conference video, created by filmmaker and Rare Dad Dan Redfield.

To date, more than 60 families and nearly 200 people have registered! Join us in Reston, Virginia, from June 1-4. Register now to attend.

If you haven't yet, **reserve your hotel room.**

For families in need of financial assistance, consider applying for a Helping Hand grant.

We are extremely grateful to the generous sponsors of this year's conference.

Presenting
Anonymous
The Doyle Foundation
Sanofi

Believe
Laura and Simeon Schindelman

Courage
Aspa Therapeutics
Chris Chapman* and William Ohle in memory of Emma Artinian

Hope
Azafaros
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In Memory of Dylan Manning and Stewart Altman
Passage Bio

Inspiration
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Blu Genes Foundation
EveryLife Foundation
Jaxson's Train for Hope
JCR Pharmaceuticals
Cameron and Hayden Lord Foundation*
Myrtelle

Family and Partners
Bonnie* and Barry Davis in memory of Adam
Codexis
Staci Kallish* and Chris Beer

Sponsors as of April 19, 2023.
*NTSAD Board Members

Email Development and Communications Director Susan Keliher to learn more about sponsorship opportunities.

Meet the Gamble Family

When Sam and Roman Gamble's son Atticus was diagnosed with Tay-Sachs disease, it felt like their world was crashing in on them. Right away, they started researching if there was anything they could do, and they found clinical trials for Tay-Sachs. Roman contacted researchers, who he found to be really nice, but they couldn't help as their clinical trials had been put on hold due to the outbreak of COVID-19.

Eventually, the initial shock of the diagnosis wore off, and Sam and Roman focused on being present and in the moment with Atticus and focused on showing him how much they loved him.

"*It's hard not to grieve, but you'll have the rest of your life for that*," Sam says. "*You've got to enjoy the time you have*."

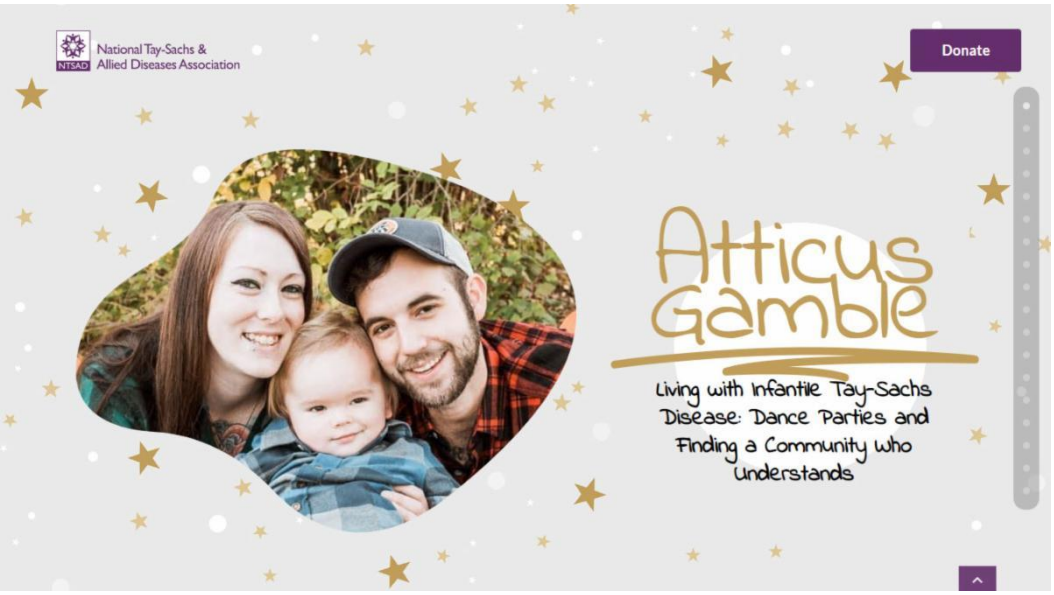
Sam and Roman had to get creative. They couldn't have friends and family over for visits because of social distancing precautions. So, they would cuddle with Atticus on a beanbag chair and look up at the colorful lights that Roman strung up throughout the house. They sometimes went for walks downtown to enjoy some fresh air.

"*We started having dance parties with Atticus every night*," says Roman. "*We'd listen to music and bounce him. He loved Russian pop music the best*."

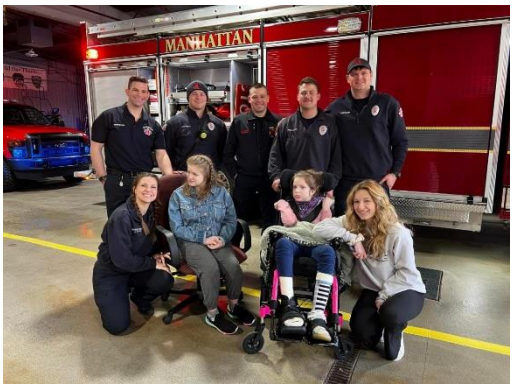
When they saw NTSAD on Facebook, they discovered a supportive and welcoming community.

"*Before we found NTSAD, it was very lonely*," says Sam. "*It was a relief to finally have people to talk to*."

The NTSAD Community provides families like the Gambles with the connection and support they need. See the impact of community by reading **The Gamble's Family's story**, and stay tuned for new stories featured each month on NTSAD's website.



The Gift of Experiences – an “Enchanted” Night for a Rare Family



Since receiving the heartbreaking diagnoses of juvenile Sandhoff disease for both daughters Mollie and Madelyn in 2016, Mandy and Jeff Ronaldson relied on their family, faith, and community. Mandy and Jeff, along with their two older children Emma and Aaron, have held annual fundraisers, raised awareness, and shared their positive outlook with other Rare Families in the NTSAD Community.

The Ronaldsons found additional support from a volunteer-led effort by their local fire department in their hometown of Manhattan, Illinois. As the family faced Madelyn’s steep decline in the loss of her abilities, the group known as **Project Fire Buddies**, stepped up with ways to make the whole family smile.

One way the Project Fire Buddies came through for the Ronaldsons was planning an incredible experience for Mollie, Madelyn, Mandy, and Emma, too—tickets to Taylor Swift’s concert in June. The seats are close enough to the stage, so they can truly enjoy the show. Before Mollie lost her ability to speak, she loved to sing, and Taylor Swift is her favorite artist.

"Mollie, she’s always been a singer," Mandy shared. "She has a really beautiful voice, she can’t sing the words anymore, but if there is music on, she’s humming, and she’s humming in tune. She and her big sister Emma—and Madelyn as well—when they were little, they would sing the 'Fearless' album, and that was Mollie’s favorite album.

It’s been a bonding experience for Emma and Mollie. They don’t have a typical sister relationship, but when they listen to that, it’s a little more typical, and they enjoy that together. It’s a reminder of what Mollie used to love, and that it’s still in there. The ‘Love Story’ song, you can see her mouthing the words, and it’s really special."

Thanks to Project Fire Buddies for recognizing that experiences like these make all the difference for Rare families.

Read the article on The Ronaldson Family and Project Fire Buddies.

If you're an affected family, **Emma's Fund for Families** at NTSAD gives grants to families to use towards enrichment activities with Rare loved ones. **Learn more.**

Give the gift of experiences, make a donation to Emma’s Fund.

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.

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