NTSAD Community News

Research, Collaboration, and Community

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

November

November 2021

In this Issue

NTSAD Welcomes New Leader

Dear NTSAD Community,

I am honored to serve as NTSAD's next leader. Having dedicated several years to healthcare advocacy organizations during three decades in the corporate, academic, and non-profit sectors, I look forward to building on NTSAD's past successes and embracing its future. With more than a dozen drug development and clinical trials underway, it is undoubtedly a time of great promise for NTSAD families. In the coming weeks, I welcome the opportunity to connect with our courageous families, Board members, industry stakeholders, researchers, and clinicians and join my voice with theirs in the fight against Tay-Sachs, Canavan, GM1, and Sandhoff diseases.

Kathleen M. Flynn, CEO, NTSAD

Read Kathy's bio.

Aspa Doses First Patient in Canavan Clinical Trial

The first participant in Aspa's Therapeutics' CANaspire clinical trial has been dosed with the company's investigational gene therapy for Canavan disease. Built on decades of research by Guangping Gao, PhD, Horae Gene Therapy Center, University of Massachusetts Medical School, this marks an important step towards bringing a potential new therapy to Canavan patients and their families.

Read the press release now.

In 2020 NTSAD interviewed Dr. Gao about his research and connection to families.

Watch the video here.

Sio Receives Fast Track Designation

Sio Gene Therapies has received Fast Track designation from the U.S. Food and Drug Administration (FDA) for AXO-AAV-GM2, the company's gene therapy for GM2 (Tay-Sachs and Sandhoff diseases). The company previously received Fast Track designation for AXO-AAV-GM1 for the treatment of GM1 gangliosidosis.

Read more.

Sio recently shared positive interim data from its clinical study of gene therapy for GM1 gangliosidosis based on UMass Chan Medical School's research.

Read more.

Fran Platt, PhD Elected Fellow of the Royal Society

Dr. Frances Platt a long-time NTSAD Community member, was recently named a Fellow of the Royal Society for her lysosomal storage diseases research. With a view to developing new drug therapies, she has identified that a treatment initially being studied as an anti-viral compound could be used to treat lysosomal diseases. Congratulations, Fran!

Read more.

Advocacy Efforts Lead to Newborn Screening Law in North Carolina

Newborn screening (NBS) is essential for early diagnosis and treatment of rare genetic diseases, like Tay-Sachs, Canavan, GM1, and Sandhoff, particularly for participation in clinical trials. To help ensure that babies born in every state have the same opportunity for early diagnosis and treatment, advocacy groups are working to pass legislation on the local level. The laws, referred to as RUSP (Recommended Uniform Screening Panel) alignment legislation, require states to screen newborn babies for any disorder on the federal RUSP panel. The laws also implement a timeline in which the screening must begin and ensure that resources will be available to fund all conditions added to the RUSP in the future.

Six states have adopted RUSP alignment legislation, including California in 2016, Florida in 2017, and Georgia, Ohio, Arizona, and now North Carolina in 2021. The current momentum of passing this potentially life-saving legislation is a result of the tireless efforts of advocates urging states to keep pace with science.

Learn more about the EveryLife Foundation's RUSP advocacy efforts.

Read the NBS Saves Lives Reauthorization Act.

NTSAD Launches New Round of Research Funding

In partnership with Blu Genes Foundation and Cure for Tay-Sachs Foundation, NTSAD will launch an RFP (request for proposal) process in January for researchers seeking seed grants ranging from $70,000 - $140,000 for one or two years, respectively.

NTSAD will be soliciting proposals for innovative research projects that involve basic research, translational studies or clinical studies in the following diseases: Tay-Sachs, Canavan, GM1, and Sandhoff diseases.

We are interested in all aspects of therapeutic discovery. Projects that focus on understanding the pathophysiology and the role of inflammation in these four disease areas, or the process of myelination and how it is affected by disease are of particular interest.

NTSAD remains committed to advancing and funding research, and first launched its Research Initiative in 2002, which to date has made more than $4 million in grants that have been leveraged to more than $30 million of additional investments in research by other institutions, leading to new therapies.

More details to come.

NTSAD Family Featured in *USA Today*

The Jackson Family—Merlie, David, and daughter Jessie, diagnosed with GM1 ganglisidosis, are beloved members of the NTSAD Community and were recently featured in a *USA Today* article that outlines how the family is living life and giving back to others. In addition, the article highlights how Jessie was one of the first patients to participate in a natural history study of the disease with Cynthia Tifft, MD, PhD at National Institutes of Health (NIH).

Jessie, who turns 30 this month, didn't get chosen for the gene therapy trial, because researchers were seeking children whose disease was in earlier stages, but the article showcases how Jessie has made a lasting impact.

"One family in the trial has three children with GM1 gangliosidosis. The dad called Merlie, asking her to thank Jessie for participating in the natural history study. **'Without her, my children wouldn't have had this shot,'** ***the father said.*** **'Tell Jessie she saved my children's lives.'**

Merlie said she 'bawled like a baby' *after that call and told Jessie,* 'You've done something the rest of us will never do.'

What more could a parent want for her child, Merlie said, **than 'to make a mark on the world and make the world a better place. Jessie's done that. So how can I be sad? She makes the world a better place every day.'"**

Read the full story here.

Support NTSAD Families

Granted: A Wish Story

Join us in supporting NTSAD Dad and filmmaker Dan Redfield, whose film, *Granted: A Wish Story*, made in honor of his daughter Ava Rose, is being featured at the 21st Annual Anchorage International Film Festival. Sadly, Ava, diagnosed with Tay-Sachs disease, passed away last week.

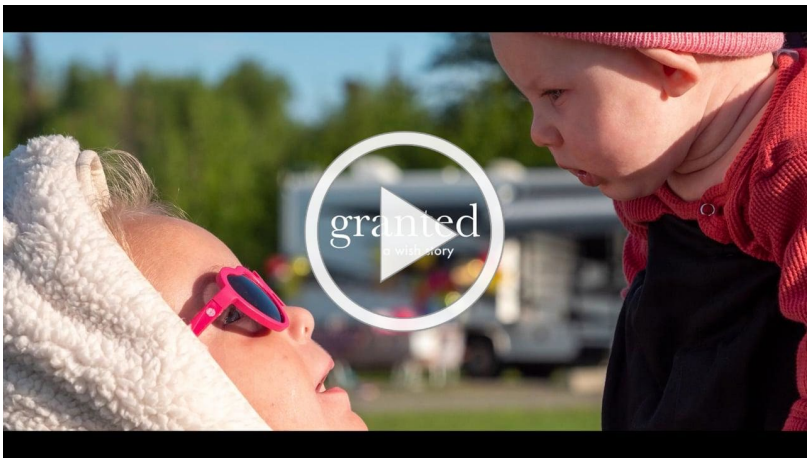
The film tells Ava's story and Dan and his wife Kristen's efforts to grant Ava and her younger sister Reagan a great adventure and lasting memories in the Alaskan wilderness.

The film's first public screening in the family's hometown is Saturday, December 11 at the Festival, and a virtual screening will be held on Friday, December 3 at 7 p.m. AST or 6 p.m. EST.

Please join NTSAD for the virtual screening and show support for Dan and Kristen as they honor Ava.

Tickets are available here.

Watch the trailer below!



Imagine & Believe!

Thank you for Imaging & Believing with NTSAD! More than 200 individuals attended the virtual event, and together we raised $120,000 in support of NTSAD Families.

Watch a video featuring NTSAD families. **Watch the full event.**

At the Imagine & Believe, we honored Sue Kahn, NTSAD's long-time executive director. Here's a thank you from Sue.

"It was a great honor to lead NTSAD for 14 years. I am proud of the extraordinary progress in research and clinical trials as well as NTSAD's ongoing efforts to put the needs of families first with comprehensive resources, programs, and services. It has been a blessing to meet so many people who have influenced me and touched my heart.

During the transition period, I have enjoyed partnering with Kathy. Thank you, Kathy and the Board for their diligence, as well as the talented staff for ensuring the ongoing care of families and driving research.

I envision a bright future for NTSAD. All things are possible for NTSAD's strong patient community and dedicated network of collaborators. There will be more tools and therapies to improve patients' daily lives. There will be FDA-approved treatments. And NTSAD will continue to be an inclusive welcoming, and cohesive patient community.

Thank you for your generosity and outpouring of kindness and heartfelt tributes during Imagine & Believe in October. I was deeply touched. We have done this important work together. Also, thank you to the families who shared the stories of their beloved children.

I wish I had the opportunity to say farewell in person to the many NTSAD families, donors, and long-time partners in industry, science, and medicine. I look forward to celebrating with you when effective treatments are found. My heart will remain with the NTSAD Community, always."

Watch our tribute to Sue.

November Marks Many Opportunities to Support NTSAD Families

November 30 is Giving Tuesday, an effort to inspire philanthropy. Consider making a gift to support NTSAD families and help sustain NTSAD's important work.

NTSAD offers programs, services, and resources for more than 750 individuals and families around the world coping with a diagnosis of Tay-Sachs, Canavan, GM1, or Sandhoff diseases. NTSAD provides a safe place where families can connect, grieve, and receive compassionate support.

"Vayle is three years old. She's a beautiful, happy little girl. Such a joy to be around. I'm so blessed to be her mom." Dawn

Dawn Mariano finds support from the NTSAD Community for her and her daughter, Vayle, who has Canavan disease.

"Being in touch with other families who know exactly what I am going through is invaluable. The disease continues to progress, and to have a network for advice —it's everything." Dawn

Make a Gift

Holiday Shopping Brings Smiles

This holiday season if you are shopping online with Amazon, consider choosing to support NTSAD as part of Amazon's charitable program, AmazonSmile. It's super easy-- whenever you shop use this link to **AmazonSmile**, and Amazon will donate to NTSAD.

Have friends and family join you in supporting NTSAD through AmazonSmile by **posting to Facebook** and **Twitter**, too!

Family Caregiver Month

November is Family Caregiver Month. Help us honor the many caregivers in the NTSAD Community by sharing our **Family Video** on social media, or by making a gift in honor of a loved one who serves as a caregiver.

Gottlieb Scholarship Recipients

For 16 years, Judy Gottlieb has been showing support of NTSAD families via the Jeffrey Alan Gottlieb and Stanley N. Gottlieb Memorial Scholarships established in May 2005 in honor of her youngest son, Jeffrey Alan Gottlieb, who succumbed to Tay-Sachs in 1975, and her husband, Stanley N. Gottlieb, who passed away in 2001. The Scholarships provide funds for college to healthy siblings in families affected by Tay-Sachs, Canavan, GM1, and Sandhoff diseases.

The 2021 Jeffrey Alan Gottlieb and Stanley N. Gottlieb Memorial Scholarship recipients are:

Matthew Kennedy, brother of Caitlin, who had Sandhoff disease, is a first-year student attending Schreiner University. Matthew is studying pre-nursing, in honor of his sister and the many nurses who cared for her.

"Through helping other people when they are ill and extending help to their family, I can return the support and kindness offered to Caitlin and my family through our difficult journey."

Gavin Levine, brother of Lila, who had Tay-Sachs disease, is a freshman in the Honors Program at Ohio State University. Gavin is pursuing a degree in biology and his interests include biotechnology and research, which he hopes to use to prevent others from losing a child.

"I envision I will use my study of biology to honor Lila's life by working to give those like her a chance to live."

Zachary Richards, brother of Cooper, who had GM-1 disease, is inspired by his brother and the nurses who cared for him. Zachary is studying nursing at Curry College.

"My dream is to become a nurse, so I can give back and work with children that have genetic disorders like Cooper...My brother lived a life that was important."

Aaron Ronaldson, the brother of Mollie and Madelyn who both have Juvenile Sandhoff disease, is a freshman at Liberty University, majoring in mechanical engineering.

"I hope to find new ways to design a lighter weight, more compact wheelchair for my sisters and others like them. I will always be thinking of them as I learn."

Thank you to Judy Gottlieb!

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

STAFF
Kathleen M. Flynn, CEO
Becky Benson, Family Services and Conference Coordinator
Sydnie Dimond, Development and Communications Associate

NTSAD
2001 Beacon Street
Suite 204
Boston, MA 02135