NTSAD Community News

Research, Collaboration, and Community

Supporting families is the center of everything we do…

June 2022

In this Issue

Dear NTSAD Community,

I am pleased to announce that our Research Initiative Program has awarded two new research projects this year. One is to study dual site application of gene therapy for GM1 gangliosidosis in felines, and the second grant funds the development of a rating scale for GM2 (Tay-Sachs and Sandhoff diseases) that enables clinicians to assess children participating in clinical trials remotely. Both of these research projects have the potential to advance research for other rare diseases. These investments in research are made possible by generous funders and individuals who support research for Tay-Sachs, Canavan, GM1, and Sandhoff diseases. Thank you to NTSAD's Research Director Valerie Greger, PhD, Scientific Advisory Committee, Research Committee, and Board of Directors for their expertise and recommendations.

Also, thanks to our sponsors and supporters, NTSAD will host our 44th Annual Family Conference in Denver, Colorado from July 7 to 10 with sessions on caregiving, family planning, living with grief, and updates on the latest research and clinical trials across all NTSAD's rare diseases. So far, more than 50 families have registered. Families can receive Helping Hand Grants to help with hotel, registration and this year, travel.

Many powerful connections are made between families at the Conference, as Rare Mom Emily so beautifully shares,

"Having a Rare child affected by these terrible diseases is an isolating experience. Being with other families at the conference and having my children be with other kids who have lost a sibling is a comforting balm. *To be with everyone and to feel the specialness of all your children, to be in company as we grieve at whatever point we are on this journey that is not confined to the life of our affected children. I look forward*

to any opportunity that helps me feel closer to my Lucy, and anything related to NTSAD does that for me." - Emily, Lucy's Mom.

Families often say that NTSAD is the family you never wanted to be a part of but you are grateful to have.

Will you give today to ensure there is a safe place for families like Emily's to receive compassionate support as they cope and heal?

We look forward to greeting Emily, her family, and everyone, including members of industry, clinicians, and researchers. All are welcome.

We hope YOU, too, will join us at the Conference!

Sincerely,

Kathleen M. Flynn
Chief Executive Officer

Support Families

NTSAD Invests in GM1 Gene Therapy Study

As gene therapy currently provides one of the most promising paths towards life-changing treatments for GM1 as well as Tay-Sachs, Canavan, and Sandhoff diseases, NTSAD has awarded a two-year grant of $140,000 to Amanda Gross, PhD, a Research Fellow at Auburn University for dual site administration of AAV gene therapy for treatment GM1 gangliosidosis in cats.

In ongoing GM1 clinical gene therapy trials the vector is either administered intravenously (IV) or in the cerebrospinal fluid (CSF). However, each of these treatment routes shows some deficiencies, and there is debate on which one is most efficacious. Dr. Gross proposes that dual-site administration (IV and CSF) will have an additive effect. The study will also advance understanding of the GM1 pathology. Results from her study, while not directly applicable to the other disorders, may nevertheless provide insights that are valuable beyond GM1 research.

Dr. Amanda Gross trained under Doug R. Martin, PhD and is currently a Research Fellow at the Scott-Ritchey Research Center, Auburn University in Alabama.

NTSAD Awards Grant for Infantile GM2 Rating Scale

NTSAD awarded a two-year grant of $139,000 to support the development of a disease-specific rating scale to be administered remotely that evaluates the status and progression of infants who have Tay-Sachs or Sandhoff disease. The grant was awarded to Elise Townsend, PhD, DPT, Michael Kiefer, DPT, and Florian Eichler, MD. The development of disease specific rating scales is a critical foundation for clinical drug trials, regulatory approvals, and the evaluation of long-term treatment for other rare pediatric genetic diseases.

"I think this tool could be a game changer in terms of clinical trials for GM2," said Dr. Townsend, an Associate Professor of Physical Therapy and Associate Director of the PhD in Rehabilitation Sciences program. ***"Use of a rating scale like the one we will develop will allow more frequent clinical assessment, without the need for families to travel to one of few care centers that provide the kind of specialized care these children need.*** *Designed to be used both in person and through virtual video conferencing, we will be able to connect with families in their homes and evaluate children's progress as they undergo experimental treatments."*

The research grant is in collaboration with the Center for Rare Neurological Diseases at Massachusetts General Hospital, and the Leukodystrophy Clinic run by Dr. Florian Eichler.

Learn more about the project.

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44 Years of NTSAD Community Coming Together

In July, the NTSAD Community will gather in-person for the first time in three years for the 44th Annual Family Conference. The four-day event from July 7 to July 10 in Denver, Colorado will bring together all types of Rare Families—those who are living with the challenges of Canavan, GM1, Sandhoff and Tay-Sachs diseases and infantile, juvenile, and late onset forms of these diseases, newly diagnosed, longtime caregivers, newly bereaved, or have lost loved ones decades ago. It's not too late to join us!

During the pandemic, the Conference held virtually in 2020 and again in 2021 connected families from more than 25 countries and had record-high attendance. Ever-resilient NTSAD families found new ways to support each other. **Supporting families is at the center of everything we do.**

Check out this year's Conference schedule.

Register today, book hotel, and learn more about the Conference here.

A look back on past Conferences:

Camp Snuggle

NSTAD's 2020 Virtual Annual Family Conference

Families Connecting at the Conference	Raleigh, North Carolina in 2019

Will YOU join us in Denver, Colorado from July 7 to 10 for the 44th Annual Family Conference?

Register Today!

Don't forget to book your hotel! **Book your hotel here.**

NTSAD offers financial assistance for families via Helping Hand Grants that can help with airfare, hotel, and travel expenses. Learn more about Helping Hand Grants.

Have questions? Don't hesitate to reach out to NTSAD's Family Services Manager Becky Benson at **becky@ntsad.org.**

Thank you to all the sponsors of the 2022 Annual Family Conference.

Support the conference by making a gift today.

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Celebrating Havi Every Day

In a beautifully written article published in the *Boston Globe,* Myra Sack, mother of Havi, shares her family's story of grief and enduring love.

“Havi comes from the Hebrew words hava and chai, both of which mean life. Havi’s middle name, Lev, translates to heart. Life and heart. We imagined Havi growing up to be a loving, generous, lighthearted person. And she was all of those things and more. They just showed up in very different ways than we ever imagined or hoped.”

When Myra and her husband Matt learned about Havi’s diagnosis that would shortly end her life, they asked themselves, *“How do we watch our daughter die? We’re supposed to just watch?”*

Uncertain what to do, it suddenly came to Matt. *“We celebrate the shit out of her. We squeeze every ounce of beauty and love into her. We decided to celebrate her with a lifetime's worth of birthdays."*

By celebrating Havi, Myra shares, ***“We were learning to live alongside grief, appreciating its power to keep us close to Havi. We were learning that pain and love could coexist. We weren’t risking our hearts, we were expanding them.”***

Myra continues, “We know the dominant narrative about grief, that it is something to ‘move beyond’ in an effort to seek ‘closure.’ But we feel differently. ***There is no safe distance when it comes to loving. Havi’s life and death has to be a part of our lives, now and forever, and we hold on to the gifts of perspective and possibility that she gave us. She exists in every moment."***

Read Myra's *Boston Globe* article.

Parent Perspective on Grief and Loss

The impact of losing a child on a parent is often misunderstood even by pediatric doctors. That's why the Pediatric Advisory Council and the National Hospice and Palliative Care Organization published an article on grief written by Becky Benson, Rare Mom of Miss Elliott and NTSAD Family Services Manager.

Read *Pediatric Journal* article on grief. (You may also right click the images of the article further below, save them to your computer, and then read the publication.)

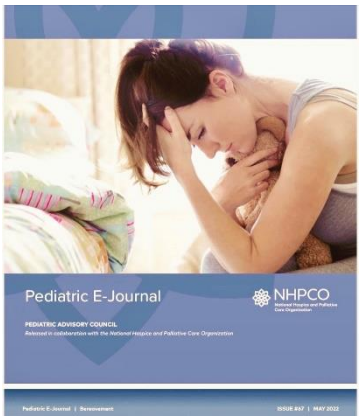
Becky’s personal and poignant art piece, *Waves of Grief,* was also recently published along with an interview. Below is an excerpt.

"People often throw around the term, 'grief comes in waves' but it's also an incredibly apt description of the overwhelming emotions one experiences. ***At times the swells feels so large they may consume you. At other moments, you find the weight of it pulling away and you're able to emerge, take a breath, and carry on.***

Usually, and for those in the long haul of grief, what seems to happen most often is that as the waves subside, you find your footing and begin to move forward when one of these waves materializes, seemingly out of nowhere and when least expected. In those moments, you can feel like you're beginning your journey all over again.

As a mother of loss, I turned to my community of fellow grievers and asked them what one thing they would want to say to their loved one who died. I included the words they shared as a part of the wave itself because even these unspoken sentiments are a part of our grief."

Becky's artwork and interview.

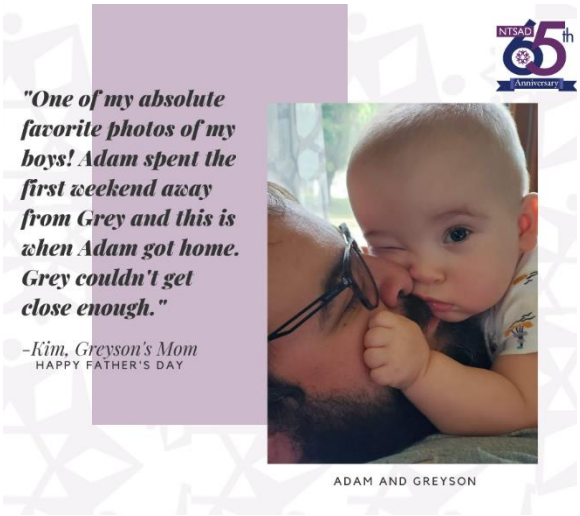


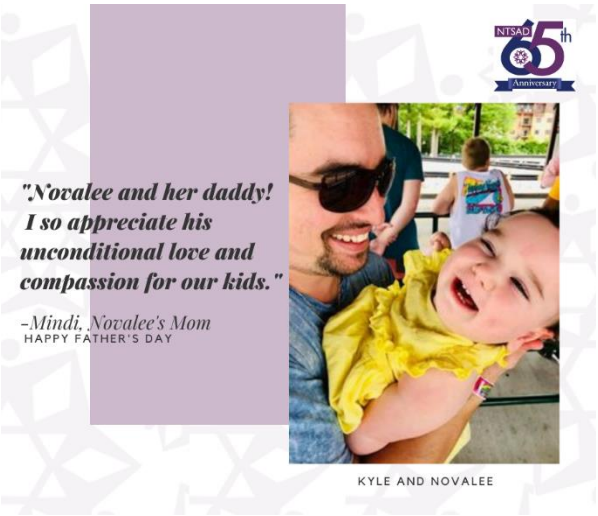
Father's Day

This Father's Day, we honor the Rare Dads of NTSAD's Community.

NTSAD recognizes the strength and enduring love of Dads who are currently caring for an affected child or adult, and of Dads grieving the loss of their child.

On Father's Day, and everyday, we celebrate the special love and bond between father and child.





GM1 & GM2 Natural History Study Webinar

Do you want to learn about the upcoming Azafaros' GM1 & GM2 Natural History Study? A webinar hosted by several GM1 and GM2 patient groups and research foundations will provide details on the upcoming PRONTO Natural History Study. Join NTSAD's Director of Family Services Diana Pangonis who works to empower patient families and who will be speaking about "the changing face of educational resources for the GM1 and GM2 community" on June 16 at 11 a.m. Eastern Time.

Register now.

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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STAFF
Kathleen M. Flynn, CEO
Becky Benson, Family Services Manager
Sydnie Dimond, Development and Communications Associate
Valerie Greger, Director of Research
Susan Keliher, Director of Development and Communications
Diana Pangonis, Director of Family Services

NTSAD
2001 Beacon Street
Suite 204
Boston, MA 02135