



# NTSAD Community News

Research, Collaboration, and Community



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

## Hello June

June 2022

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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](#)

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## 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.

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## 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



NTSAD'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](mailto: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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Martha\* and Dr. Ron  
Kleinman

\*NTSAD Board Member

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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.](#)

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## 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.)

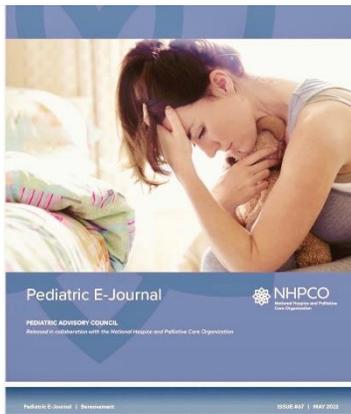
Becky's personal and poignant art piece, *Waves of Grief*, was also recently published along with an interview. Below is an excerpt.

*"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.



Pediatric E-Journal | Bereavement | ISSUE #17 | MAY 2022

### Bereavement, a Parent's Perspective

Becky A. Benson  
Editor's note:  
Family Services Manager  
National Tay-Sachs & Allied Diseases Association  
Wilmington, NC

With *Phenylketonuria* (PKU) being added to the DSM for the first time, redefining Persistent Complex Bereavement Disorder, bereavement has more officially become pathologized. Persistent Grief Disorder does not have the usual diagnostic features such as persistent, intrusive, distressing, or anxiety, or severely, prolonged and disruptive for the parent/guardian for the parent who has lost and only persisting at least three of the eight symptoms listed here, one year after a final event's death.

**These include:**

- Feeling as though part of you has died
- A sense of disbelief about the death
- Involvement of memories that the parent is dead
- Strong emotional pain related to the death (anger, bitterness, or sorrow)
- Difficulty moving on with your life (socializing with friends, pursuing interests, planning for the future)
- Emotional numbness
- Feeling that life is meaningless
- Extreme loneliness (feeling alone or separate from others)

But what most parents who have lost an child already know is that many if not most of these so-called symptoms are normal and exhibited to some extent by most parents specifically, and that for many, their grief is too intense to let go of so easily. Many parents would agree that there is no timeline to their coming through or beyond their grief. It is an ongoing and fluid. When someone has told me, we will sleep now then, for the rest of our lives, it's a natural and part of the human experience. It poses the question, that what if grief cannot be described? And for bereaved parents, it could be better in your mind and that it's incredibly difficult to describe as the fact that it often is both unspoken and unspoken. It's not certain that many of us feel we have lost a part of ourselves in a very real and true sense.

Additionally, for those wanting to be medically taught or formally to child, your daily routine and all aspects of life are impacted in your life with your child. It's not surprising that when a child dies, these parents may also feel a loss of their sense of purpose, or even that sense of self. When parents may combine with feelings of helplessness, simply because they were unable to save their child. As a parent this can lead to feelings of both guilt and shame as they perceived their responsibility for their child or even suspect that they, themselves, and self-compassion play an important role in a parent's ability to move forward. As this stage encompasses when the individual feels one space experiencing great pain is already completely equipped. I don't know a single parent of two who didn't find themselves to be somewhat numb at some point in their grief.

Pediatric E-Journal | Bereavement | ISSUE #17 | MAY 2022

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**Issue #17 | Bereavement**

**Bereavement, A Parent's Perspective**  
Becky A. Benson

This article is a meditation on grief, its expression, and the value of support. The article concludes with these thoughts: "The process of mourning is not one that can be said should be rushed, it cannot be bypassed for the sake of society's comfort. It should not be suppressed or ignored. We should be actively working to create a community of grievers parents, in order to support them through their grief no matter how long it lasts because after all, grief is the cost of a great love lost."

Pediatric E-Journal | Bereavement | ISSUE #17 | MAY 2022

While the symptoms of grief may become harmful, such as the inability to interact with the world around you, grieving has been, and may still be, a process of becoming completely compassionate and empathetic toward the health of the world. It is important to recognize the ways in which these and other symptoms and prevent it from becoming a source of suffering. It is important to recognize that there are children who still need a diagnosis of grief to help them understand the pain they are experiencing. It is important to recognize that there are children who still need a diagnosis of grief to help them understand the pain they are experiencing. It is important to recognize that there are children who still need a diagnosis of grief to help them understand the pain they are experiencing.

Bereaved parents want to desperately to find a way to continue to work in what feels for them, a world broken into two parts: the before and after. Parents desire to be able to show and talk about their child with others, meet the way those who have longed to be able to do. For the bereaved, their children are still very present in their thoughts. In the heart of it all, bereaved parents may feel that as the world around them moves forward, it will forget about their child completely. One should not shy away from mourning their child's name to a grieving parent. It is worth to their own. Additionally, they may struggle to move forward in terms of the uncertainty of what they mean they are leaving their child behind. While this is, of course, not the case, it does take time for the bereaved to modify their response to the new world without their loved one and adjust to living in it.

Support is never better in a grieving parent's ability to move forward. There will never be the final closure for the initial impact. This reaction, but may feel feelings of isolation, anger, or hurtful when these initial supports fall away. Many parents report that feeling responsible for the initial cause of their child's death, sorrow, and unrelenting pain. Most wish the outside world didn't expect them to suddenly be OK after a period of unrelenting and arbitrary time, such as an or a week or months. They wish their friends and family would just check in on them and recognize this will always be a permanent scar on their face.

The experience of community has been a constant factor for me throughout my daughter's diagnosis, illness, and past her death, making a meaningful support group of others who understand firsthand and a safe space for my grief. It has been very hard to lose my daughter's death, and it's still in my mind, light and heavy. I find my advice to, with those I can talk to, but my wife, them, and there are vulnerable moments. The grievers who have lost their child report sharing this feeling on.

The process of mourning is not one that can be said should be rushed, it cannot be bypassed for the sake of society's comfort. It should not be suppressed or ignored. We should be actively working to create a community of grievers parents, in order to support them through their grief no matter how long it lasts because after all, grief is the cost of a great love lost.

## 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.

**NTSAD 65<sup>th</sup> Anniversary**

*"Èl es quien hace que todo sea posible. admiro profundamente a este hombre. Lucho te amo."*

*"He is the one who makes everything possible. I deeply admire this man. Lucho, I love you."*

*-Alejandra, Facundo and Guiliiana's Mom*  
HAPPY FATHER'S DAY



LUCHO Y SU FAMILIA

**NTSAD 65<sup>th</sup> Anniversary**

*"One of my absolute favorite photos of my boys! Adam spent the first weekend away from Grey and this is when Adam got home. Grey couldn't get close enough."*



*-Kim, Greyson's Mom*  
HAPPY FATHER'S DAY

ADAM AND GREYSON

**NTSAD 65<sup>th</sup> Anniversary**

*"Novalee and her daddy! I so appreciate his unconditional love and compassion for our kids."*



*-Mindi, Novalee's Mom*  
HAPPY FATHER'S DAY

KYLE AND NOVALEE

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## 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.](#)

## WHAT IS A NATURAL HISTORY STUDY?

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Join us to hear about the Azafaros PRONTO Natural History Study and the innovative educational resources created by Cognitait to support patients in the study.

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**16 JUNE 2022**

8 am (PT) / 11 am (ET) / 4 pm (GMT) / 5 pm (CET)

**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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