2023 IMPACT REPORT

Bringing Our Community Together

NTSAD

National Tay-Sachs & Allied Diseases Association

REMEMBRANCE

In memory of NTSAD community members who passed from July 1, 2022, through June 30, 2023.

IN MEMORIAM

William Leo Bursak

Nevaeh Cunningham

Riley Fennaughty

Elliott Greenberg

Frederick (Fred) Horak

Nayelie Jean-Baptiste

Siena Maria Margani

Victoria Rocha

Greyson Rudness

Mallory Rayne Salazar

Paulina Barros de Castro Waldman

Richard Wallack

Annabella Wright

We sincerely apologize for any unintentional omissions from this list.

> “There is no masking of suffering, and there’s no hiding or protecting from it. When the world tells you, ‘Don’t bring your suffering to me,’ the NTSAD Community says, ‘You’re welcome here.”
>
> **SARA SCAPAROTTI**
> MOM TO JOEY AND
> NTSAD BOARD VICE PRESIDENT

Meeting the Needs of Our Community

Dear Families and Friends of NTSAD,

You, your engagement, and financial contributions support children, adults, and families coping with a diagnosis and loss from Tay-Sachs, Canavan, GM1, and Sandhoff diseases. We continue to fight until these devastating diseases are no longer fatal or debilitating.

NTSAD has a 66-year history of responding to our community’s needs by creating new programs and resources, expanding advocacy initiatives, and broadening our research efforts to adapt to the shifting landscape of these rare diseases.

Since its founding in 2002, the Research Initiative has awarded more than $4.7 million in 72 research grants. Without such an investment, the advancements in drug discovery and clinical trials would not have been possible.

In recent years, we have been helping individuals and families navigate clinical trials by sharing information and making connections and supporting them as they cope with disappointment and grief if they do not qualify. No matter where a person or family is in their rare journey, we are here to help.

We listen to our community. We ask questions, solicit opinions and feedback, and create a program or resources to meet unmet needs. Sometimes we bring in outside speakers or specialists to provide additional support or expertise. We initiate opportunities to raise awareness and empower our community to share their experiences with each other, researchers, industry, and the U.S. Food and Drug Administration (FDA). In this Impact Report, we highlight some of the many events and resources we created during the past year.

Thank you for supporting families and investing in our efforts to accelerate research. Together, we have made incredible strides. You make our work possible.

With gratitude and determination,

Oralea Marquardt, LCSW
Mother to William
Board President

Kathleen M. Flynn
Chief Executive Officer
NTSAD

To learn more about the impact of your giving, please visit our new website: ntsad.org.

Our Community in Action

On March 21, 2023, several parents whose children are or were affected with Canavan disease shared their experiences during a Patient-Led Listening Session with the FDA, so its members could learn more about the disease. Thank you to all the parents who participated. For more, please visit page 5.

Family Services

174 attendees | **49** families

Attended the 44th Annual Family Conference in **Denver, Colorado**, July 2022

305 attendees | **78** families

Attended the 45th Annual Family Conference in **Reston, Virginia**, June 2023

22 attendees | **11** families

Attended a Regional Family Meetup in **Irvine, California**

18 attendees | **8** families

Attended a Regional Family Meetup in **Austin, Texas**

814

Individuals and families connected through the Family Support Group **Facebook page**

13

Individuals and families reached out for initial support via the **NTSAD website** since its launch in February 2023

~1,000

People received **Lifeline, a monthly newsletter** for families

~20

Families with newly diagnosed family members and newly bereaved families received specialized **family connections monthly newsletters** for a year

12

Monthly Zoom sessions were held to help adults with Late Onset Tay-Sachs or Sandhoff connect and support one another

Family Services

Meeting the Needs of Families

NTSAD's dedicated Family Services Team of Diana Jussila (formerly Pangonis) and Becky Benson connect with families every day. To provide rare families with opportunities to support one another, have meaningful conversations, and form local friendships, the team created new regional Family Meetups. The first was held in Irvine, California, in September 2022, and a second in Austin, Texas, in February 2023.

Together Again!

After two years of holding Annual Family Conferences virtually during the pandemic, NTSAD hosted two in-person Annual Family Conferences.

The 44th Annual Family Conference was in Denver, Colorado, from July 7 to 10, 2022, and 174 people attended. Nearly $50,000 in Helping Hand grants were awarded to 28 families who would have been unable to attend without financial support.

More than 300 people participated in the 45th Annual Family Conference in Reston, Virginia, from June 1 to 4, 2023. A record 28 children affected by rare diseases attended. Helping Hand grants totaling $90,000 were awarded.

Special thanks to an anonymous donor and the Doyle Family Foundation for providing funds for Helping Hand grants to the 2022 and 2023 Annual Family Conferences.

NTSAD Is a Safe Place for Everyone

At the Annual Family Conference in Virginia, several Spanish-speaking families attended from Argentina, Chile, and Mexico. The National Institutes of Health provided free translation services. Luis and Alejandra, founders of Cutasa, a patient advocacy group in Argentina, shared information they received at the Conference with more than 50 families.

Bringing the Patient Voice to the FDA

On March 21, 2023, five parents of children with Canavan disease, as well as a physical therapist, shared their experiences during a first-ever Patient-Led Listening Session for Canavan disease with the U.S. Food and Drug Administration (FDA). The purpose of the meeting was to educate FDA members on symptoms, diagnosis, and complex daily care. A report summarizing the meeting is on NTSAD's website. This listening session was a collaborative effort by NTSAD, The Canavan Foundation, and Cure Canavan Fund.

Research

$4.7 MILLION
has been awarded in grants since 2002

72
Grants have been awarded to **47 scientists** since NTSAD's Research Initiative began in 2002

$375,000
given to research projects in FY23.

Driving Research Is at the Core of Our Mission

Research Initiative

This year marked the second year since the re-launch of NTSAD's Research Initiative, after a strategic review of focus areas, which include basic science and translational research, clinical development, earlier symptomatic diagnosis, and newborn screening.

During the second grant cycle, NTSAD awarded financial grants to three research projects that focus on tools that support future studies and trials, including the development of severity scales for disease staging, translational biomarkers with clinical utility, and measurable and meaningful efficacy endpoints for new therapeutic approaches. The grants will provide $170,000 in funding to the three projects over two years. The projects include:

Amanda Nagy, MD, and Florian Eichler, MD

Project: Characterization of Progressive Neuroimaging and Pathologic Changes in Canavan Disease*

Jennifer Kwon, MD, and Julie Kissel, PhD Candidate

Project: Development of a Disease-Specific Clinical Rating Scale for the Late-Onset GM2 Gangliosidoses

Dominic Gessler, MD, and Guangping Gao, PhD

Project: Non-Invasive MRI-based Therapeutic Outcome Prediction Modeling Using Machine Learning*

**NTSAD is grateful to The Canavan Foundation and the Jonathan and Amy Greenberg Fund at NTSAD for their partnership and support of these grants.*

NTSAD Supports University of Massachusetts Chan Medical School GM2 Trial

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

> ***"We 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, Mathew Forbes Romer Foundation, Cure Tay-Sachs Foundation, and Blu Genes Foundation for providing the funding that has enabled this trial to continue."***

NTSAD contributed $80,000, and the total amount given to this project including contributions from Mathew Forbes Romer Foundation, Cure Tay-Sachs Foundation, and Blu Genes Foundation was $320,000.

More Research Accomplishments

- NTSAD provided second-year funding to Elise Townsend, DPT, PhD, PCS, from Massachusetts General Hospital for Constructing and Validating an Infantile GM2 Rating Scale and to Amanda Gross, PhD, from Auburn University for Dual Site Administration of AAV Gene Therapy for the Treatment of Feline GM1 Gangliosidosis.
- Researchers and clinicians attended a special Scientific Symposium at the 2023 Annual Family Conference: Translating Preclinical CRISPR Editing in Tay-Sachs and Sandhoff Rodent Models to Human Clinical Trials.
- NTSAD launched its first-ever virtual biorepository of patient samples to facilitate the sharing of cell lines and other patient samples among researchers and clinicians. NTSAD serves as a connection point between researchers but does not physically collect or house the samples.
- The Katie & Allie Buryk Fund sponsored the 5th Annual Late Onset Tay-Sachs and Sandhoff (LOTSS) Think Tank retreat to bring together clinicians and scientists and propel research and potential treatments for LOTSS.

Partnering with Industry

Representatives from biotechnology and biopharmaceutical companies, along with patient advocacy leaders and members of academia, participated in the inaugural meeting of NTSAD's Industry Roundtable at the 2022 Annual Family Conference. The goal of the Roundtable is to forge collaboration and encourage efficiencies among participants. A key accomplishment was the creation of Industry Partnership Guidelines.

Special Thanks to Donors for Research Staff Support

Cameron and Hayden Lord Foundation
Katie & Allie Buryk Research Fund
Mathew Forbes Romer Foundation
New York Area Fund
Sussman Family Fund
Vera Pesotchinsky Research Fund

Development and Communications

6,451
People received *Community News*, NTSAD's e-newsletter

1,950
Gifts were made to support families, advocacy, and research

11,000
Visitors came to the new NTSAD website since it was launched in February 2023

942
gifts totaling
$283,486
were made to the Annual Fund

15%
increase
over last year

25+
individuals participated in the Day of Hope, which raised nearly $50,000 for critical research

SOCIAL MEDIA FOLLOWERS

3,300 Facebook
620 X (Twitter)
691 Instagram
555 LinkedIn
92 YouTube

(Figures as of June 30, 2023)

Development and Communications

Creating Resources for Families and Our Community

For the first time in a dozen years, NTSAD launched a new website with a focus on families. The new website, **ntsad.org**, highlights families, their stories, their photos, and their experiences. Knowing that many first-time visitors to our website are individuals and parents who recently received a devastating diagnosis, our goal is to provide easy-to-understand language about care and symptom management, the latest research, clinical trials, and the support our community provides.

We sincerely thank the following families for sharing their stories: Samantha (Sam), Roman, and Atticus Gamble; Merlie, David, and Jessie Jackson; Dawn and Vayle Mariano; Mona and Owen Vogel; and Stephanie and Joey Wright.

Commemorating NTSAD's 65th Anniversary

In November 2022, and for the first time in three years, the NTSAD community gathered in person for *Imagine & Believe*, our signature fundraising event. At the event, we honored Guangping Gao, PhD, for his extraordinary efforts to revitalize gene therapy and identify the Aspa gene for Canavan disease, and his work in developing potential gene therapy treatments. To read more about Dr. Gao, see page 11.

Every Day is a Day of Hope

Since 2011, families have hosted events and activities to honor and remember their loved ones and raise awareness and funds for research. In recent years, members of industry and rare allies have participated as well. This year, we created a unifying event to encourage everyone's participation. Some walked, rolled, strolled, or even drove a mile for hope—and we held an online gathering for our community to commemorate Day of Hope, together. Thank you to families and the greater community for raising nearly $50,000 for research.

Thank you to the following donors who sustain NTSAD by making monthly or recurring gifts to support our mission.

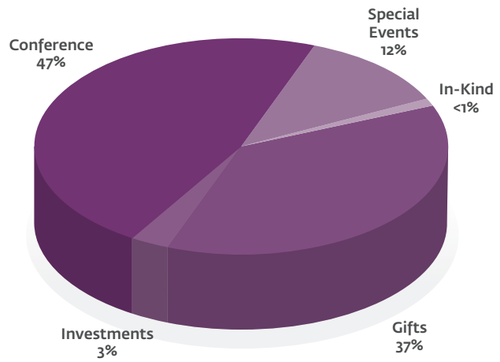
Marcia Cohen
Wit Koenig
Laura Kowal
Gray Levine
Sarah Ohle
Matt Steckman
Angela and Leon Tynes
Ryan Wilichinsky

If you would like to set up a recurring gift to NTSAD, please contact the Development Team at 617-277-4463 or visit ntsad.org.

Financial Information

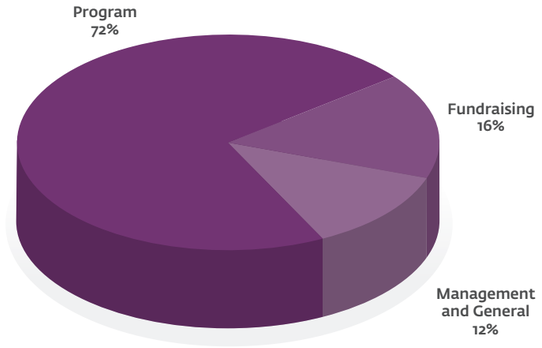
REVENUE

Gifts	$445,566
Conference	$554,854
Special Events	$139,287
Investments	$37,785
In-Kind	$8,721
Total Revenue	**$1,186,213**



EXPENSES

Program	$1,084,315
Fundraising	$246,629
Management and General	$185,583
Total Expenses	**$1,516,527**



NET ASSETS AT THE BEGINNING OF THE YEAR: **$3,091,561**

NET ASSETS AT THE END OF THE YEAR: **$2,772,520**

As a result of NTSAD's recent relaunch of its Research Initiative, $384,930 in prior years' donor restricted funds supported research projects in FY2023. This planned expense resulted in a negative change of $319,041 in total net assets. However, NTSAD continues to maintain a strong fiscal position, as evidenced by a $65,889 positive change in total unrestricted net assets.

Special Recognition to Members of the NTSAD Community

We thank and acknowledge the countless individuals who give their energy, commitment, and expertise to support families and drive research. In 2022-2023, we recognized the following individuals for their extraordinary accomplishments and contributions to the NTSAD community.

Guangping Gao, PhD

NTSAD 65th Anniversary and 2022 *Imagine & Believe* Honoree

Known as the godfather of gene therapy, Dr. Guangping Gao is the Co-Director, Li Weibo Institute for Rare Diseases Research, Director, Horae Gene Therapy Center and Viral Vector Core, Professor of Microbiology and Physiological Systems, Penelope Booth Rockwell Professor in Biomedical Research, University of Massachusetts (UMass) Chan Medical School. Dr. Gao began studying Canavan disease in 1989. He developed a Canavan gene therapy in 2017. His work was licensed by Aspa Therapeutics, where clinical trials have begun.

Mimi Blitzer, PhD, and Erin Strovel, PhD

2023 Above and Beyond Awardees

Each year at our Annual Family Conference, NTSAD recognizes an individual or a group who are not directly affected by a rare disease, and yet have served our community. This year, NTSAD awarded Mimi Blitzer, PhD, and Erin Strovel, PhD, for their efforts that led to the completion of NTSAD's Tay-Sachs Testing Quality Control Program.

Risa Asnen

2022 Above and Beyond Awardee

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.

Holly Stringer, Mom of Brennan

2023 Power of One Awardee

Every year at NTSAD's Annual Family Conference, we recognize someone who has been affected by a rare disease and supports other rare families through personal service. In June 2023, Holly Stringer, mom to Brennan, was recognized for creating the Conference's Camp Activities Program for affected kids.

Daniel and Patricia Lewi

2022 Power of One Awardees

Daniel and Patricia Lewi established the Cure and Action for Tay-Sachs Foundation (CATS Foundation) in June 2011, after their daughter Amelie was diagnosed with Tay-Sachs. At the time, there was no such charity based in the United Kingdom (UK).

National Tay-Sachs & Allied Diseases Association

National Tay-Sachs & Allied Diseases Association
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EIN# 13-1912877

Follow Us

Our Vision

We envision a world in which Tay-Sachs, Canavan, GM1, and Sandhoff diseases are no longer fatal or debilitating. To uphold our vision, NTSAD will:

- Provide compassionate support, advocate for affected individuals and their families, and promote early diagnosis and prevention
- Empower, educate, and connect affected individuals and families
- Be a key resource for individuals, families, clinicians, researchers, and industry members
- Direct, promote, and invest in research to accelerate the development of treatments, therapies, and cures
- Serve as a leader within the rare disease community

NTSAD Mission

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.