NTSAD

National Tay-Sachs & Allied Diseases Association

60th ANNIVERSARY
1957–2017

NTSAD recognized its 60th anniversary throughout 2017 by honoring those families and individuals who established and expanded the NTSAD community, by recognizing our present achievements, and by embracing goals for the future in the areas of family support, research advances, and preventative outreach and education.

Spotlight on **Imagine & Believe**

NTSAD celebrated its 60th Anniversary Imagine & Believe event at the Royal Sonesta in Cambridge on Thursday, November 9th. Highlights of the night included a thoughtful discussion with honorees Dr. David Meeker and Professor Timothy Cox. Moderated by NTSAD's former Executive Director, Jayne Gershkowitz, topics included what motivates their work in the rare disease community, scientific progressions, and the value that an organization like NTSAD can have on both families and supporters. This discussion was followed by heartfelt remarks by Talia's mother and NTSAD parent, Carla Steckman.

Joining Carla and honorees, David and Tim, were 190 guests who **raised over $120,000—the event's largest total since its inception!**

NTSADResearch

Translating Patient Experience to Help Research

NTSAD partnered with Canavan Research Illinois and Cure GM1 Foundation on Patient Insights Networks (PIN) for Canavan and GM1 diseases. The PIN's purpose is to allow families to enter information about their disease experience that is translated into deidentified data to paint a picture of the disease experience across a global community. The data will ultimately help promising research advance to clinical trials by bolstering the hard data of the research.

NTSAD and Cure Tay-Sachs Foundation (CTSF) have partnered to launch a GM2 Tay-Sachs and Sandhoff Disease Patient Insights Network (PIN) this spring.

Million Dollar Bike Rides Raise $40,000 for NTSAD Grant

Team NTSAD raised $40,000 from the Fourth Annual Million Dollar Bike Ride held in Philadelphia, and the amount was matched by UPenn's Orphan Disease Center! The 2017 grant went to Igor Nestrasil, MD, PhD, at the University of Minnesota for his project "Brain MRI signatures in infants with infantile forms of GM-1 and GM-2."

Progress in Gene Therapy Research

The Tay-Sachs Gene Therapy Consortium completed animal studies and the follow-up pathology studies. The results met the criteria to advance to the next stage. They will next manufacture clinical grade vectors with a goal in 2018 to submit an Investigational New Drug (IND) filing to start clinical trials.

2017 Research Initiative Grants Awarded

- Proof of concept study of HSC gene therapy for Tay-Sachs disease

  Principal Investigator: Alessandra Biffi, MD
  Dana-Farber/Boston Children's Cancer and Blood Disorders Center

- Minimally invasive delivery of AAV gene therapy in the Tay-Sachs Sheep

  Principal Investigator: Heather Gray-Edwards, PhD
  Scott-Ritchey Research Center, Auburn University, Auburn, AL

- Development of a quantitative method for the determination of a pentasaccharide in GM1-gangliosidosis patient cells to assess the potential therapeutic efficacy of a beta-galactosidase pharmacological chaperone drug candidate.

  Principal Investigators: Tim Wood, PhD, Greenwood Genetic Center, Greenwood, South Carolina and Stephane Demotz, PhD, Dorphan, Lausanne, Switzerland

- Accelerated program for CSF delivery of AAV gene therapy for Tay-Sachs and Sandhoff patients

  Principal Investigator: Miguel Sena-Esteves, PhD
  University of Massachusetts Medical School

NTSADEvents

Day of Hope

2017 Day of Hope: Families Drive Research

- Over $101,000 raised, bringing the total to **over $320,000 since 2011**
- More than 20 events and over a dozen custom t-shirt campaigns
- Families rallied and raised funds for research from coast to coast including Canada

Community Rallies around Family

We would like to give a special thanks to the Ronaldson family and friends for **raising over $50,000 for research** with their Day of Hope event. Mollie and Madelyn, who have Juvenile Sandhoff, are an extraordinary inspiration to not only their friend and friends, but to families worldwide.

NTSADFamily

NTSAD's ANNUAL FAMILY CONFERENCE: A Community of Support

The Annual Family Conference is the cornerstone of NTSAD's Family Services program. Families impacted by all forms of Tay-Sachs, Sandhoff, GM1 and Canavan diseases come together to support and empower each other through similar and shared experiences. The program encompasses all facets of living with these diseases from peer support, research updates, care management and choices, to a focus on relationships, sibling support and how to be an advocate.

3 FULL DAYS

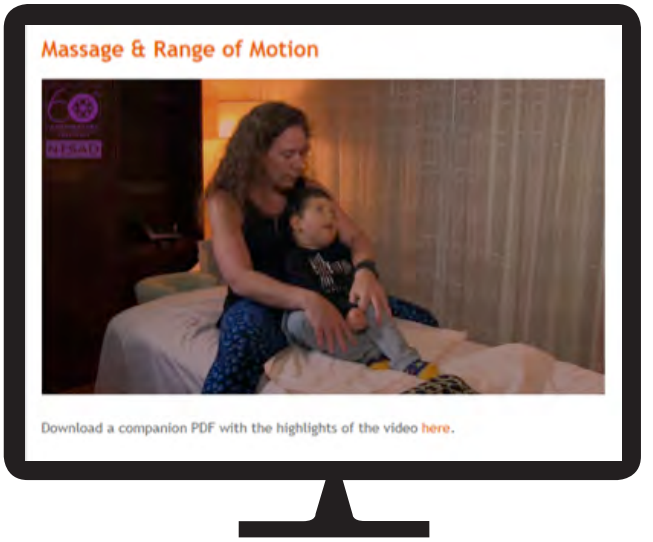
OVER $50,000 IN HELPING HANDS GRANTS given to help families attend the **2017 Annual Family Conference**

40+ FAMILIES HELPED

New Support Resources

Care Tip Series for Families

The first two videos of the series launched in the summer of 2017 to educate families on issues relating to caring for a child. The topics covered "G-Tubes A to Z" and "Motion & Massage." The series will expand in the summer of 2018 to include two more topics relating to care.



Emma's Fund for Families

This new fund, started by the Artinian family in memory of their daughter, Emma, was established to allow affected families to apply for grants to use towards enrichment activities. It can be used any way a family feels would be beneficial for creating their memories, such as trips to the zoo, photo shoots, tickets to an event, an overnight in a hotel with a pool, etc.

NTSADAdvocacy

NTSAD, with inspiration and guidance from the EveryLife Foundation and the National Organization for Rare Disorders, continued to encourage families to reach out to their representatives throughout the year. Sharing their stories can stress the importance of voting for legislation that can enable families to be whole and healthy.

InRemembrance

Jason Arbetter
Seaby Clark
Jeter DeJong
Ava Demeshko
Cristian Fernandez
Avi Forrester
Cash Gray
Kylie Hatchel
Amelie Lewi
Levi Lucero, Jr.
Cora McDonald
Jenna McKinzie
Alyssa Mushin
Declan O'Connell
Ian Ortega
Rion Page
Mackenzie Plick
Barron Posey
Cooper Richards
Josiah Scaparotti
Talia Steckman
Nikko Zollar

January 1, 2017 – December 31, 2017

NTSAD**Awareness**

Expanded Carrier Screening

Progress was made in carrier screening through the Counsyl–JScreen research project, The Hexosaminidase A Variants of Unknown Significance (HAVUS). The goal of this project was to reclassify common HEXA VUS to likely benign/benign, thereby conferring a higher carrier detection rate for sequencing. This research will be a meaningful milestone in the evolution for Tay-Sachs carrier screening from enzyme analysis to sequencing.

This project was funded by NTSAD and affiliate family foundations.

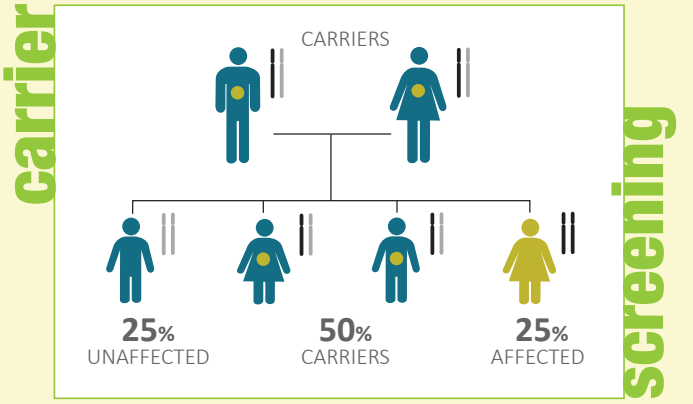
KNOW THE FACTS:

one in 27 Ashkenazi Jews, French-Canadians, and Cajuns...

one in 50 to 1 in 190 Irish-Americans...

one in 250 of the general population...

... is a carrier of the Tay-Sachs gene.

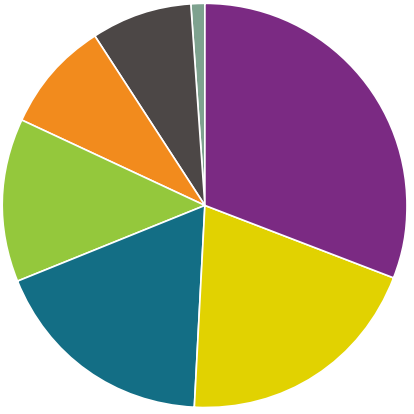


NTSAD encourages meeting with a genetic counselor to understand your risks before getting tested.

2017**Fiscal Year**

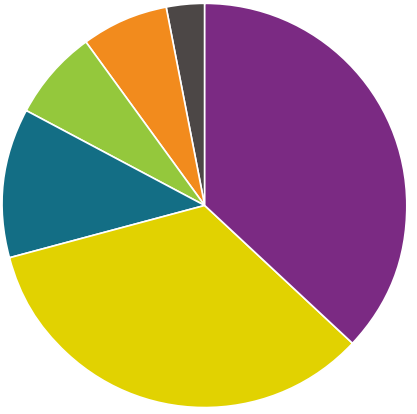
Audited Financial Statements

SUPPORT & REVENUE
$809,635



- **31%**—Research
- **20%**—Gifts
- **18%**—Conference
- **13%**—Special Events
- **9%**—Grants
- **8%**—Restricted Gifts
- **1%**—Other

EXPENSES
$711,679



- **37%**—Research
- **34%**—Family Services
- **12%**—Fundraising
- **7%**—Education
- **7%**—Administration
- **3%**—Advocacy

NTSAD**Mission**

NTSAD leads the fight to treat and cure Tay-Sachs, Sandhoff, GM1, Canavan and related genetic diseases, and supports affected families and individuals in leading fuller lives.

National Tay-Sachs &
Allied Diseases Association

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