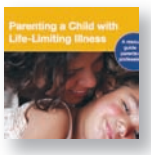
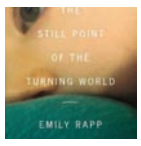
National Tay-Sachs and Allied Diseases Association SPRING 2013

KEEPING UP WITH NTSAD

Second Annual Day of Hope exceeds expectations
Raising funds for research remains top priority for families.



New NTSAD film released this spring
A compelling DVD resource available for families to lessen isolation after diagnosis.



The Still Point of the Turning World
NTSAD mom's recently released book brings nationwide attention to Tay-Sachs.

World Rare Disease Day
NTSAD represented at World Rare Disease Day observances in 2012 and 2013.

NTSAD Celebrates 55th Anniversary with Two Galas

Over 300 of NTSAD's friends and family members gathered in Boston on November 1 and in the New York area on December 5 to celebrate NTSAD's 55th Anniversary.

Shari Ungerleider, Emil Kakkis, NTSAD honoree, Sue Kahn and Kevin Romer (left to right)

Former CEO of Genzyme, Henri Termeer, was honored in Boston for his pioneer role in bringing treatments to people with rare diseases. He spoke about his motivation to develop treatments because "you never give up once you look into the eyes of a patient." Emil Kakkis, MD, PhD, founder and president of the EveryLife Foundation, was honored at our New York Gala for his dedication to improving the lives of those living with rare diseases. He offered encouragement about the impressive Tay-Sachs gene therapy progress and made a generous gift to encourage others to help fund "those last few yards to the finish line."

The benefit events raised over $160,000 in support of NTSAD's mission to treat and cure Tay-Sachs, Canavan and related genetic diseases, and support affected families and individuals in leading fuller lives. NTSAD parents, and sister-in-law and brother-in-law, Blyth Lord and Tim Lord, spoke about the value NTSAD brings to families. Each affirmed the importance of supporting "our small but mighty" NTSAD family as we honor the tradition of "friends taking care of friends, generation after generation."

Henri Termeer, NTSAD honoree

PRESIDENT'S LETTER

Dear Friends,

As I prepare to pass the torch to the next NTSAD Board President, I must share with you how proud I am of the work our NTSAD community has accomplished in the last year. Highlights include: celebration of our 55th anniversary with successful galas in Boston and New York, exceeding our fundraising goal for the Second Annual Day of Hope, holding our biggest family conference in Orlando, and the premiere of a new film to help families cope with their child's diagnosis.

NTSAD continues to drive and manage the progress and challenges toward major research breakthroughs giving unprecedented hope to those diagnosed with these diseases. Your invaluable support of NTSAD's mission and our families makes it all possible.

My sincerest thanks,

Kevin Romer
NTSAD Board President

Hope was raised across the country from Florida (Kyla left), to Texas (Ashley right).

9.22.2012- Second Annual Day of Hope Exceeded All Expectations

On September 22, 2012, NTSAD's Second Annual Day of Hope, families across the country raised over $47,000 for research. They hosted events big and small ranging from community walks to neighborhood lemonade stands. The top fund-raiser, the Fernandez family, of Miami, Florida, won the opportunity to meet with Miguel Sena-Esteves, PhD, Principal Investigator of the Tay-Sachs Gene Therapy Consortium. Plans are in motion for the Third Annual Day of Hope which will be held on September 21, 2013.

NTSAD Research Initiative Update

The Research Initiative Grants awarded in 2012 focused on advancing the pre-clinical work of the Tay-Sachs Gene Therapy Consortium with an eye toward initiating clinical trials.

Doug Martin, PhD Auburn University
1. Sheep as a Model of Tay-Sachs (Year 3)
2. Supplemental Pre-Clinical Studies of AAV Gene Therapy in Feline Sandhoff Disease

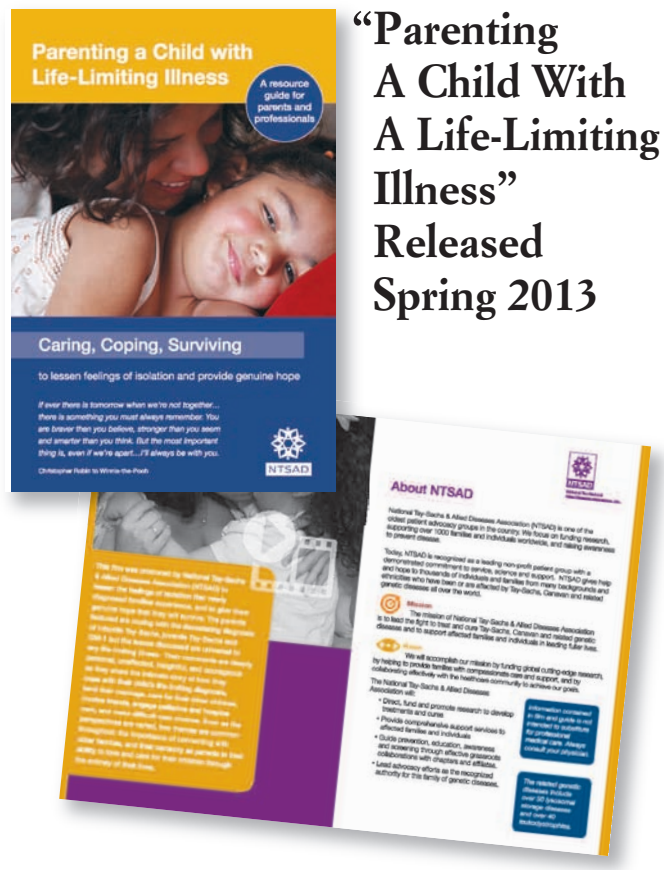
St. Jude's Farm / Fred and Joan Horak
For ongoing of care and feeding of affected Jacob sheep flock

Thomas J. Conlon, PhD University of Florida
Supplemental equipment for toxicology studies

University of California, Davis
(co-funded with Cure Tay-Sachs Foundation)
GMP manufacturing of AAV vector for Tay-Sachs gene therapy clinical trial

Nine other Research Initiative grants were completed in 2012 or are in progress. Progress reports are available through the *Research We Fund* section of our website and the website's library. The projects completed include: Canavan gene therapy, biomarkers, induced pluripotent stem (IPS) cell therapy, anti-inflammatory therapy, and high throughput screening assays. Over $2.5 million has been awarded over the last 12 years leading to over $10 million in NIH funding toward finding a cure.

"Parenting A Child With A Life-Limiting Illness" Released Spring 2013



NTSAD has produced and released, "Parenting a Child with a Life-Limiting Illness," a new film and resource for families designed to lessen the feelings of isolation they experience when their child is newly diagnosed. Five NTSAD families discuss everything from coping with their child's diagnosis and caring for their marriage, to tough end-of-life decisions. This project was funded with a Patient Advocacy Leadership grant award from Genzyme - a Sanofi company.

The universal themes of coping and surviving will resonate with all parents of terminally children, reaching beyond the NTSAD community. The film will also be a teaching tool for health care providers to improve understanding of the parent perspective and help them provide more thoughtful and supportive care to the entire family. To learn more and to see a trailer, visit www.NTSAD.org.

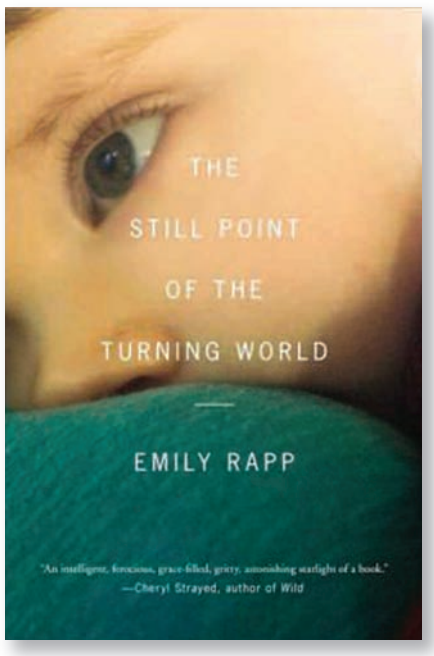
NTSAD Launches *Lifeline Online* For Families

In October 2012, NTSAD launched a monthly online version of its powerful family newsletter, *Lifeline.* Like its quarterly cousin, the mission of *Lifeline Online* is to foster a community of support and empowerment for families affected by Tay-Sachs, Sandhoff, GM-1, Canavan and allied diseases. This monthly electronic newsletter focuses on topics such as health management, coping and connecting families.

World Rare Disease Day

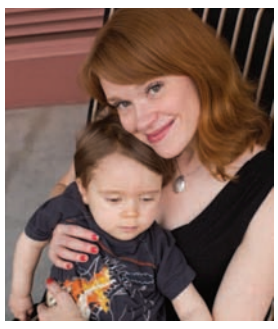
The last day of February is known as World Rare Disease Day. It is an international advocacy day to bring widespread recognition of rare diseases as a global health challenge. Nearly 7,000 rare diseases affect roughly 30 million Americans. NTSAD is proud to have been a part of the 2012 and 2013 World Rare Disease Day observances at the Whitehead Institute in Cambridge, Massachusetts, the State House in Boston, and at the New Jersey State House in Trenton.

Rare Disease Day



"The Still Point Of The Turning World"

NTSAD parent, Emily Rapp's poignant, moving and raw book about her experience parenting a child with Tay-Sachs, was released on March 7th. Her unflinching honesty and eloquent words in the essays and columns inspired the book, and as a result have captured the hearts and minds of many loyal readers. In addition to her appearances on NBC's "The Today Show," and NPR's "Fresh Air,"excerpts of her book have been featured in The Boston Globe, L.A. Times, New York Times, Oprah Magazine, Time and Vogue. Visit www.NTSAD.org to learn more.



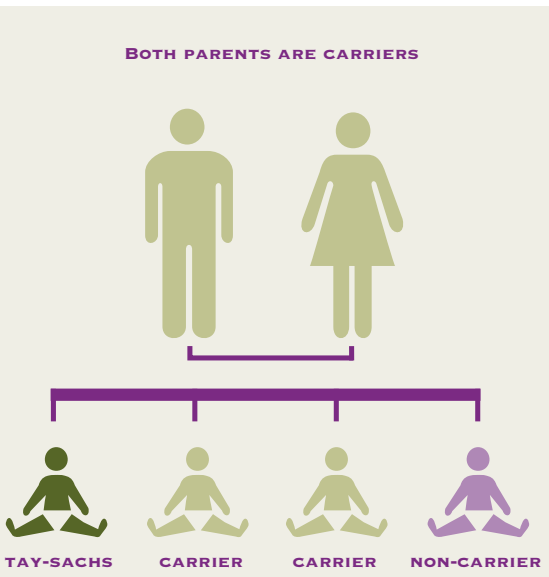
Emily's son, Ronan, peacefully passed away on February 15th surrounded by family and friends. He was almost three years old.

The More You Know…

1 in 27 Ashkenazi Jews, French-Canadians, and Cajuns; 1 in 50 to 1 in 190 Irish-Americans; and 1 in 250 of the general population is a Tay-Sachs carrier.

NTSAD recommends enzyme testing as the primary method for identifying Tay-Sachs carriers. The enzyme test is a simple blood test. DNA testing typically detects only the most common Ashkenazi Jewish gene mutations. **Always** consult a genetic counselor for guidance.

While enzyme testing remains the gold standard in Tay-Sachs carrier screening, NTSAD continues to support the development of newer and more accurate methods of carrier screening. For example, NTSAD families have donated blood samples, which have been a helpful resource for companies studying the accuracy of new testing technologies.



If you have questions or concerns about carrier screening, please call NTSAD at (800) 906-8723.

Keep Up with NTSAD! Receive the latest news and updates via e-mail! Send your email and any address changes to **Ingrid@ntsad.org** and we will be sure to keep you up-to-date on all our news!

NTSAD's spending is consistent with its mission. In fiscal year 2012, program expenses accounted for 84% of total spending, in line with non-profit best practices. Almost 50% was spent on Research programs and 25% was spent on Family Services. More financial information and NTSAD's audited financial statements are available at www.NTSAD.org.

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