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National Tay-Sachs & Allied Diseases Association, Inc.

**Keeping Up with NTSAD**  
a monthly e-newsletter

### August 2014 - In This Issue:

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**TOGETHER.**  
**We can make a difference.**

## NTSAD In the News

*The Jewish Daily Forward* publishes their annual Genetics issue every summer which is always informative and at the same raises awareness. They recently published two stories featuring members of the NTSAD family in their annual Genetics issue. These articles are an example of the ripple effects NTSAD has on families throughout their respective journeys. In addition they published an ad for NTSAD in the print edition of *The Forward* that emphasizes the importance of carrier screening.

Genetics

### A Network for Courageous Parents Offers New Views, Advice on Grief

*Having Company Helps in Worst of Times*



In Memoriam: Oralea Marquardt, pictured with her family, holds a photo of her son William, who died of a rare genetic disease in February 2014. She talks about her experiences in a video for the Courageous Parents Network to ease the sense of isolation for other parents.

Oralea and Rod Marquardt with their children, William, Kyla and John. William passed away from GM-1 in February this year.

Genetics

### Two Grieving Mothers, One Friendship

*How Two Women From Different Backgrounds Became 'Grief Mentors'*



A Friendship Built on Grief: Emily Rapp (left, with her son Ronan) and Becky Benson (with a picture of her son Elliott) became friends after their children were diagnosed with the fatal Tay-Sachs disease.

Emily, on the left, with her son, Ronan. Becky, on the right, holding a picture of her daughter, Elliott.

One story (left) focuses on NTSAD parent Blyth Lord's **Courageous Parents Network**, a site

for parents whose children have received a terminal diagnosis of any kind.

The other article (right) is about the close friendship between two NTSAD mothers, Becky Benson and Emily Rapp who bonded by their experiences as mothers of children who had Tay-Sachs.

Read these articles online [here](#).

## RESEARCH

### Three Year Study of Jacob Sheep Complete

Dr. Douglas Martin and his team of researchers at Auburn University have recently completed a three year study on sheep with Tay-Sachs disease (TSD). In addition to further characterizing this disease model, Martin's group conducted safety and efficacy studies of AAV gene therapy in the TSD sheep. Since the sheep have a mutation in the HEXA gene (and subsequent deficiency of Hexosaminidase A (HexA) protein), this is an authentic model of Tay-Sachs disease. Furthermore, the large brain size of the sheep allows researchers to confront certain challenges of gene therapy, including brain targeting and distribution.



### What Did They Learn?

They learned that survival rates increased after TSD sheep were treated. The most successfully treated group had a 57% increase in lifespan. They also found that enzyme levels were highest in the areas of injection in the brain with it decreasing with more distance from injection site. The same was found with the reduction in ganglioside storage. The MRIs of the treated sheep indicated improvement after AAV gene therapy.

### What Happens Now?

The one gene therapy treated sheep that is still alive and doing well at almost 1.5 years of age will continue to be monitored regularly. Dr. Martin and his team will also continue to optimize gene therapy in the sheep model of TSD, primarily focusing on better distribution of

the enzyme to more distal brain regions (cerebellum) and the spinal cord.



We are pleased to say that the veterinarians at Auburn University had a very successful first breeding season with 13 lambs already born and two more expected in the near future. These lambs need names and can be adopted and named with a donation of at least \$1,000. Please contact Joan Lawrence at [joan@ntsad.org](mailto:joan@ntsad.org) to adopt and name one of the lambs, or you can make a gift online [here](#).

We hope that you will support the effort to sustain this flock of Jacob Sheep which are a valuable animal model of Tay-Sachs disease. Thanks to our generous donors over the last five years, NTSAD has made grants totaling over \$300,000 for sheep research studies and for maintaining the Jacob sheep flock.

## CLINICAL RESEARCH NETWORK

On July 10th, members of NTSAD's Scientific Advisory Committee, clinicians from industry involved in rare diseases, researchers, a systems expert, and NTSAD representatives gathered at a **GM2 Clinical Research Network** meeting in Boston at Massachusetts General Hospital. While the meeting focused on GM2, the experts gathered are knowledgeable about Tay-Sachs, Sandhoff, Canavan, and GM1, so the discussion applied to all of our diseases. All are interested in working together to expedite clinical trials for GM2 and related diseases.



To read the original article featured in an NTSAD **Topic of the Week**, click [here](#).

## POSTDOCTORAL FELLOWSHIP OPPORTUNITY



On August 4th, the University of Pennsylvania's Center for Orphan Disease Research and Therapy (CODRT) announced a [request for applications \(RFA\)](#) for a \$35,000 Postdoctoral Fellowship in Tay-Sachs, Sandhoff, GM-1, Canavan and related diseases. This opportunity extends the impact NTSAD's Research Initiative has on research in the field of these rare genetic diseases. To further the impact, gifts for research can be made [here](#).

The funding results from money raised from the [Million Dollar Bike Ride](#) in Philadelphia on May 3rd, 2014. *Note: Next year's bike ride is May 9, 2015.*

## EDUCATION, AWARENESS & ADVOCACY

### NTSAD Parents on Capitol Hill

In late July, a group of NTSAD moms went to Capitol Hill to advocate for expanded palliative care services. The initiative was called the Palliative Care Lobby Day and it was organized by the [Patient Quality of Life Coalition](#). One of our parents, Michelle, wrote about her experience in a recent *Topic of the Week*. She highlighted, "Since the [July] 23rd, a number of cosponsors have stepped forward to support both pieces of legislation and we hope there will be enough support to move forward with these measures in the months to come, but as everyone in Washington, D.C. knows, this is an election year and successfully moving legislation forward right now, even when it has broad support, is a challenging business."



To read her article in its entirety, click [here](#).

## MEET STEW.

An amazing, intelligent and quick-witted man. A husband, brother, uncle, father and grandfather. He is incredibly proud of his beautiful grandchildren. He prides himself on his loyalty to the Yankees. Stew has also been living with [Late Onset Tay-Sachs \(LOTS\)](#) for more than a few decades now. He was a young man when symptoms started and progressively worsened over the years. All the while though Stew keeps on living. He works. He travels to conferences with his loving wife, Lorrie, albeit not every year as traveling is more stressful now. He serves on the NTSAD Board of Directors offering his insight and experience on a number of issues, and stays in touch with others affected by LOTS. He and Lorrie are invaluable members of the NTSAD family as they guide others in their journeys.



## UPCOMING NTSAD HAPPENINGS

NTSAD's work is made possible thanks to the support of our annual events and gifts to the Annual Fund. NTSAD solely relies on the generosity and philanthropy of private gifts and grants.

### [Secret Poker Run and Ride for Hope](#)

**Saturday, August 23, 2014**

**The Sandbar, Akron, Ohio**

The Secret Family will host their first annual poker run and ride this Saturday in honor of their daughter, Lily, to raise funds for Canavan research. If you

cannot make it to their outing, consider making a gift to support their efforts [here](#).

**Family Fun Walk in honor of Danny Kenny**  
**Sunday, September 7, 2014**  
**Pearl River, New York**

The Whitney and Kenny families will host a walk/run in honor of Danny Kenny who is fighting his battle with Tay-Sachs. They are working with NTSAD's former New York area chapter members to raise awareness and funds for research. At the same time there will be an Irish Carrier Screening Study held from 11am to 2pm conducted by the Einstein College of Medicine funded in part by NTSAD's Delaware Valley Chapter in Philadelphia. Click here to learn [more](#).



\$15 per rider  
\$5 per passenger  
\$20 per car

First bike out @ 11:00am  
Last bike in @ 5:00pm

**POKERBUT**

**AUGUST 23, 2014**

**STARTS AND ENDS @ 5:00**

**THE SANDBAR**  
3822 S. MAIN ST  
AKRON, OHIO

Food  
Entertainment  
Fun

Silent auctions  
Raffles

NTSAD

EVENTS FOR HOPE

**Sixth Annual 'Fore' Jack Memorial Golf & Tennis Outing**  
**Wednesday, September 17, 2014**  
**Piping Rock Club, Locust Valley, NY**

The Klinger family will host their sixth golf and tennis outing on Wednesday, September 17th in memory of their son, Jack, whose valiant battle with Canavan disease ended when he was 14 years old. To take part in this wonderful event, contact Joan Lawrence, Development Director, at [joan@ntsad.org](mailto:joan@ntsad.org).



**Fourth Annual Day of Hope, September 20, 2014**

It is an NTSAD tradition now to raise and rally funds for research in the months leading up to September every year. This day gives families, their friends and community the opportunity to shout their children's names, raise awareness and fundraise for research. Activities range from lemonade stands and bake sales to community walks and spaghetti dinners. Our goal is to raise at least \$50,000 this year for research. To see if something is happening near you or to get ideas, visit the NTSAD Events page [here](#).



If you'd like to do some kind of activity or donate to support this special day, visit [www.NTSAD.org](http://www.NTSAD.org) for more information.

**7th Annual Boston Benefit: Imagine & Believe**  
**Thursday, November 13, 2014**  
**Royal Sonesta Hotel, Cambridge, MA**

The date has been set for another fantastic evening celebrating NTSAD's work. It will take place on Thursday, November 13th at the Royal Sonesta Hotel in Cambridge, MA. We are thrilled to honor [Stephen Groft, Pharm.D.](#), former head of the Office of Rare Disease Research at the National Institutes of Health in Washington, D.C.



To sponsor this special evening, purchase an ad in the program book or buy tickets, contact Joan Lawrence, Development Director, at [joan@ntsad.org](mailto:joan@ntsad.org) or (617) 277-4463.



I want to give NTSAD  
my support today.

Give a gift here.

Sue Kahn, Executive Director ([skahn@ntsad.org](mailto:skahn@ntsad.org))

Joan Lawrence, Development Director ([joan@ntsad.org](mailto:joan@ntsad.org))  
#204

Ingrid Miller, Office Manager ([ingrid@ntsad.org](mailto:ingrid@ntsad.org))

Diana Pangonis, Communications Manager ([diana@ntsad.org](mailto:diana@ntsad.org))

SACHS

NATIONAL TAY-

& Allied Diseases Association,

2001 Beacon St -

Boston, MA 02135

(617) 277-4463 \* [www.NTSAD.org](http://www.NTSAD.org)

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