

Lifeline Online

July/August 2015
NTSAD Family Newsletter



NATIONAL TAY-SACHS &
Allied Diseases Association



NTSAD

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& allied diseases association

www.NTSAD.org

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electronic publication of
NTSAD for families.

Information contained in
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consult your Physician.

Dear Families,

This edition of "Lifeline Online" is a new format covering all the topics that relate to our small but mighty group. It is more of an online magazine! Because these issues are for you, I would love to hear your thoughts, comments, suggestions and always welcome content. If you like to write and would be interested in helping with the topics that are covered in these monthly issues, let me know! I would love to have one or more volunteers to help with the following topics:

- Health Management & Equipment
- Late Onset Community Issues & Resources
- Juvenile & Canavan related Issues & Resources (Schools, IEPs)
- Births & Memorials
- Kids' Corner (for siblings)
- Research Topics
- Advocacy Actions & Alerts

If you're interested, please let me know sooner than later! Give me a call at (800) 906-8723 or email me at diana@ntsad.org.

In the meantime, enjoy the remaining days of summer and the days leading up to the Day of Hope!

Diana

Calling All Siblings of All Ages!

We want to start a **Kids' Corner** newsletter but we need contributors and an editor to help collect all the amazing articles, stories, poems and pictures that the siblings will submit!

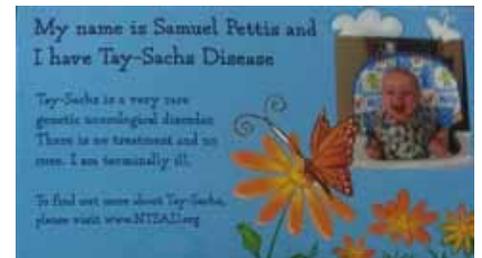
Contact Diana at diana@ntsad.org if you're interested in volunteering as the Kids' Corner editor!

SUPPORT

Explaining Your Condition or Your Child's Condition to Strangers

There have been a few posts in the NTSAD Family Support Group page asking about cards to give to strangers to educate them about your or your child's disease. Not only do you get to share your story and answer their inevitable questions, but you get to raise awareness! Several families have printed their own using various websites such as GotPrint.com or Vistaprint.com.

NTSAD also has cards for our Late Onset friends. Let Diana (diana@ntsad.org) know if you'd like some send to you.



The Rare Siblings Project

(www.raresiblingsproject.org) is a collaboration between NTSAD and Courageous Parents Network thanks to the generous Patient Advocacy Leadership award from Genzyme - a Sanofi company.



The project is for siblings, by siblings:

- Siblings sharing their experience to help other siblings

It is also for parents, by parents:

- Parents sharing their perspective to help other parents who wonder and worry about the challenges of caring for all their children.

The site will grow and the resources will expand. If you have questions, let Diana know at diana@ntsad.org.

Jeffrey & Ira Gottlieb Scholarships Due ASAP

If you are a sibling, in your first, second, third or fourth year of school, submit your application as soon as possible. Email Diana at diana@ntsad.org to receive the guidelines.

A Facebook Question with Advice from Other Parents



How do you find the right nurses?

Make sure you let the [agency] know you need an RN with experience. Set and be clear with your expectations.

Remember...the nurses and their agencies work for you and your loved one.

Start with day nursing so you can show them how you like to do things.

Make a list of non-negotiable qualities of nurses who take care of [your child].

If you need more information regarding this topic, let Diana know or post in the NTSAD Family Support Group page.

LATE ONSET CORNER

Scott Hunger
Joins
NTSAD Board
of Directors!



I am a down-to-earth guy who is an attorney and MBA but tries not to be limited because I also happen to have been diagnosed with Late-Onset Tay Sachs in the year 2000. I currently use a motorized wheelchair during the waking hours. I am very interested in advocating on behalf of NTSAD with national politicians and I have the skills to assist NTSAD in reaching and then surpassing its financial goals. I would also like to use my professional network to help NTSAD become nationally supported by the public and hence, by politicians. I also maintain a full-time professional position as a Contract Attorney and enjoy writing, hanging out with friends, talking on the telephone and watching TV and movies in my spare time.

Get in touch with Scott at sahunger@verizon.net.



**Three Ways to
Connect
without
Leaving the
Comfort of
Home**

These popular services are a great way to connect to the rest of the Late Onset community throughout the year. Another place to post your news, questions or comments is on the Late Onset Facebook page. Keep the conversation going!

Published Paper Addresses Pyrimethamine

In 2009, NTSAD funded an investigator-sponsored clinical trial at Hospital for Sick Children and NYU for the treatment of late-onset with pyrimethamine (PMT) led by Drs. Joe Clarke and Ed Kolodny. The results were published in 2011. (An open-label Phase I/II clinical trial of pyrimethamine for the treatment of patients).

The study concluded that leukocyte Hex A activity is enhanced in vivo by treatment with PMT. However, future studies were needed to assess the protocol, including dosage, and perform related biochemical studies. An Israeli team of researchers performed similar studies and recently published their results.

While they also found that PMT increases Hex A activity in LOTS patients, "the observed increase is repeatedly transient and not associated with discernible beneficial neurological or psychiatric effects."

Visit www.NTSAD.org to download the paper from the NTSAD Library.

Join the Late Onset
Facebook Group.

Late Onset Tay-Sachs
a closed group
www.facebook.com

ADVOCACY

21st Century Cures Passed in the House

July 10, 2016, Rare Disease Legislative Advocates (RDLA) Blog: Historic Vote in House Passes 21st Century Cures and the OPEN ACT with 344 ayes, 77 noes. We applaud Chairman Upton, Representative DeGette, House Leadership and all the rare disease advocates who took action to help pass this lifesaving legislation.



The 21st Century Cures Initiative: This landmark legislation will spur scientific discovery and bio-medical innovation, providing hope for patients and their families, while also protecting jobs and America's leadership role in healthcare innovation.

Attention now turns to the Senate, and we look forward to working with Senators on their ideas and priorities for creating treatments and cures for rare diseases. We encourage the Senate to move quickly in advancing this legislation through the chamber and to the president's desk.

Stay tuned to NTSAD's Facebook page and emails for future updates on this groundbreaking legislation!

NTSAD Toolkit for Meeting with Your Representatives

Thanks to Stephanie Bozarth and the National MPS Society, we've created a packet of information that can be customized for your meeting with any state representative.

It includes:

- NTSAD "A Common Bond" Booklet
- NTSAD Annual Reports
- Information about the disease that impacts your family
- A talking points sheet to help with what is usually a highly organized meeting
- A summary of current legislation affecting our rare disease community



UNITED STATES
HOUSE of
REPRESENTATIVES

To find out who your state representatives are in your area, visit www.house.gov/representatives/find/.

RESEARCH

A CALL TO ACTION FOR FAMILIES...

We need information from you that will ultimately help us provide the correct data to researchers. Oftentimes, researchers and pharmaceutical companies want to know the answers to critical questions in order to decide whether they should pursue a potential treatment or conduct further studies. If possible we want to collect the answers to these questions:

1. What was the date of diagnosis?
2. Where was the diagnosis made?
3. When was your loved one born?
4. When did your loved one succumb to the disease?



While we may already have the information, it would be a great help to have you send it again. Email Diana at diana@ntsad.org for the form or download it from the library at www.NTSAD.org under Family Support.

Thanks!

NTSAD Funded Research in 2015

Over \$266,000 in grants were awarded this spring to five research projects. The grants were made for a one to two year period for up to \$40,000 a year. These projects represent NTSAD's commitment to fund research that will lead to treatments for rare genetic diseases such as Tay-Sachs, Canavan, GM1 and Sandhoff diseases.

Grants were made for the following proposals:

Development and Validation of a Rapid MS/MS-based Method to Detect HexA deficiency in Tay-Sachs disease

Principal Investigator: Denis Lehotay, PhD
University of Saskatchewan College of Medicine

Intravascular Gene Therapy for Feline GM2

Principal Investigator: Douglas Martin, PhD
Auburn University

Generation of a Knock-in Mutant Hexb Mouse Model

Principal Investigator: Eric Sjoberg
OrPhi Therapeutics

Late Onset Registry and Repository

Principal Investigator: Florian Eichler, MD
Massachusetts General Hospital

Defining Natural History of Canavan Disease through the Development of an International Registry

Co-funded with The Canavan Foundation
Principal Investigator: Heather Lau, MD, MS
Co-Investigator: Paola Leone, PhD
New York University

Do you receive the monthly emailed Research Reviews from NTSAD? If not, email Ingrid@ntsad.org to put your name on the list.

Join the Fifth Annual Day of Hope on September 19th, 2015!

It is our time to make a difference, to rally and raise funds for research. You can do anything on that day to take part - host a lemonade stand, have a yard sale with the neighbors, post a giving link to support Day of Hope on your Facebook page and encourage your friends to do the same, or organize a community walk/run. Really anything will have an impact.

To receive a Day of Hope care package, email Ingrid at ingrid@ntsad.org and let her know what you need.

HOPE 
knows no boundaries

www.NTSAD.org

#LipSync4ACure



Thanks to Sherri Manning, we may have our version (a much drier one) of the Ice-Bucket Challenge.

Here are the rules:

1. Pick your favorite song.
2. Challenge three friends to do the same or make a gift to NTSAD.
3. Record a 30 second clip of you lip-syncing to it and making the challenge.
4. Lip-sync with all your heart for all those who cannot sing!
5. Share on Facebook #LipSync4aCure.

Let's see this take off. It's been already reported that the ALS Ice Bucket Challenge raised enough to see a breakthrough happen in ALS research. We can do the same!

Giving Pages to Make Life Easier for You

NTSAD can now host your own giving page that you can share with friends, family and the community. The Day of Hope is a great time to start one, but we can create one for any occasion that is meaningful to you.

All you need to do is send Diana (diana@ntsad.org) a few of your favorite pictures, a message to share and set a goal. From there we can track gifts on a thermometer that is displayed on your personal page! It's that easy. Visit the 2015 Day of Hope page at www.ntsad.org to see other pages!

SEEKING WRITERS, CONTRIBUTORS, GUEST COLUMNISTS FOR LIFELINE!

If you have a few minutes to spare, like to write, have ideas for future issues and want to contribute to "Lifeline" please let Diana know at (617) 277-4463 or diana@ntsad.org. We want to be sure these monthly issues are helpful to everyone. All perspectives are welcome - parents, siblings, grandparents, caregivers and our Late Onset friends!



Summer...NTSAD Family Style

Someone once said, "Summer should get a speeding ticket." How true. As the unofficial end of summer approaches, we get to relive some favorite and blessed summer moments on this page.

If you have a favorite picture of summer, feel free to share it on the NTSAD Family Support Group page on Facebook!





In memory

The NTSAD Family has had to recently say goodbye to their loved ones. Please take a moment, read their names aloud and hug those you hold dear in their honor.



Samuel Pettis
Received his wings on
August 25, 2015



Meghan Cicolini
Received her wings on
August 21, 2015



Colby 'CB' Chappell
Received his wings on
August 13, 2015



Amelie Joyce
Received her wings on
July 20, 2015

Make music with what remains.
Complete the song left for us to sing,
Transcend the loss,
Play it out with heart, soul and might
With all remaining strength within us.

- Rabbi Harold M. Schulweis