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NTSAD

National Tay-Sachs &
Allied Diseases Association, Inc.

Keeping Up with NTSAD
a monthly e-newsletter

October 2014 - In This Issue:

TOGETHER.
We can make a difference.

IMAGINE & BELIEVE 2014

This fun Boston benefit for NTSAD will take place on **Thursday, November 13th**. Last year's sold-out event raised over $80,000! **RSVP online here** to help us surpass last year's results and join us for a fantastic evening! *The deadline is*

Monday, November 3rd.

Royal Sonesta Hotel, Cambridge
November 13th * 6:00-9:30pm

NTSAD is happy to honor **Stephen C. Groft, PharmD**, former Director of the Office of Rare Diseases Research at NIH. His work advocating for the rare disease community has given hope to thousands of families and individuals affected by rare diseases. It will be a special evening as friends, families and members of the rare disease community come together to honor Steve, NTSAD's work and each other's amazing support of our small, but mighty family.

If you or someone you know would like to attend on **Thursday, November 13th** please contact Joan Lawrence at **joan@ntsad.org**. You can also **RSVP online here**. Gifts can also be made in honor of this special evening through our website **here**.

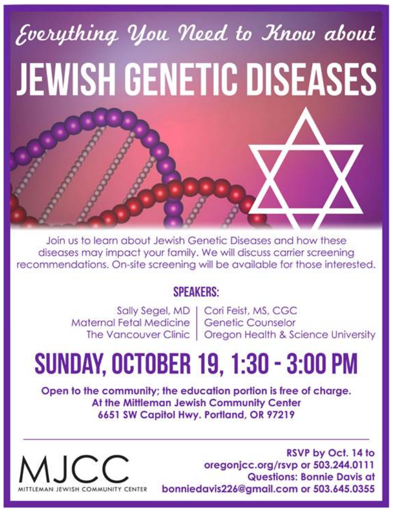
EDUCATION, AWARENESS & ADVOCACY

A Bar Mitzvah Project and A Screening Program

NTSAD parent Bonnie Davis organized a community screening and educational event at her local Jewish community center in Portland, Oregon on Sunday, October 19th. However, the day began at her son's Sunday school where he spoke about his mitzvah project in front of the whole religious school.

"For my bar mitzvah project, I am supporting National Tay-Sachs & Allied Diseases Association. I am passionate about this because my older brother, Adam, died from Tay-Sachs, which is a Jewish genetic disease. I never knew my brother personally; he died before I was born. However, his life has had a great impact on my family."

Jeremy raising awareness and funds for the NTSAD Helping Hand Grant Fund!



Everything You Need to Know about

JEWISH GENETIC DISEASES

Join us to learn about Jewish Genetic Diseases and how these diseases may impact your family. We will discuss carrier screening recommendations. On-site screening will be available for those interested.

SPEAKERS:

Sally Segel, MD
Maternal Fetal Medicine
The Vancouver Clinic

Cori Feist, MS, CGC
Genetic Counselor
Oregon Health & Science University

SUNDAY, OCTOBER 19, 1:30 - 3:00 PM

Open to the community; the education portion is free of charge.
At the Mittleman Jewish Community Center
6651 SW Capitol Hwy. Portland, OR 97219

MJCC
MITTLEMAN JEWISH COMMUNITY CENTER

RSVP by Oct. 14 to
oregonjcc.org/rsvp or 503.244.0111
Questions: Bonnie Davis at
bonniedavis226@gmail.com or 503.645.0355

After Jeremy's presentation, they moved to the community center where a few dozen people attended the program. The audience heard from a gentleman affected by a rare disease and learned about the diseases from a local physician and genetic counselor from the Oregon Health & Science University. Ten people attending were screened that day and interestingly there were a number of young adults from a group called the Moishe House which is a social group for young Jewish adults, post-college and pre-marriage.

Hope for Change in Washington, D.C.

NTSAD mom, Sherri Epstein, attended the BIO Patient and Health Advocacy Summit in Washington, D.C. on behalf of NTSAD. In attendance were other patient groups as well as representatives from Capitol Hill, the FDA and the NIH.

Sherri reflected on how good it was to connect with other rare disease groups and know that pursuing treatments for rare diseases requires banding together to affect real change. She also felt reassured to hear that lawmakers understand the importance of special waivers to allow families struggling with rare diseases have access to drugs that can ease the progression of the diseases in their loved ones.

While funding research is one step to helping children and adults affected by rare diseases, it is also important to make sure that policies are in place to allow access to those future treatments. Collaborating and connecting with other rare disease groups will undoubtedly give a stronger voice to those who need to be heard.

NTSAD thanks Shire for granting a travel stipend to enable Sherri to attend the summit. To learn how you can be involved with advocating like Sherri and raising awareness like Bonnie, please contact the NTSAD office at (617) 277-4463.

After a great conference on how to get through the paper work to get a cure for rare diseases, I'm so motivated to go full speed ahead with conquering Canavan Disease!!

RESEARCH

Progress: The Name of the Game

NTSAD's Executive Director, Sue Kahn, attended the **National Organization for Rare Disorders (NORD) 2014 Breakthrough Summit** this week. The Summit convened the top leaders from the FDA, NIH, Industry, Patient Groups, Payers and Research Institutions to address the progress of rare disease diagnosis, genomics, drug development, patient engagement, product approvals, FDA oversight and market accessibility to orphan products.

One central theme throughout the two-day summit was that the patient voice is an important part of the drug development and treatment process. Janet Woodcock, MD, Director of the FDA's Center for Drug Evaluation and Research said, "For the past 30 to 40 years, patient groups have focused on funding research; that fertilizes the soil. However, it's not enough to make the vegetables grow." She outlined several ways patient groups can be engaged that can have better outcomes and a higher return on investment.

1. What is the current knowledge of the disease(s)?
2. What is the natural history of the disease(s)?
3. What causes the disease(s)?
4. What is the current state of existing therapies/interventions?

The answers to these questions can only be answered with the involvement of families affected by the diseases along with guidance from NTSAD's Scientific Advisory Committee (SAC) and Corporate Advisory Council (CAC), and that is why it is so important to NTSAD to be the conduit for that information. For example, last week a secure link to update family information was published; if you have not done that and would like to, **please use this link** here.

NTSAD is on the Right Track

Last week's e-published "Topic of the Week" featured NTSAD's monthly "ResearchReview" and highlighted several projects that are examples of how NTSAD's community is engaged in the ways Dr. Woodcock mentioned above.

To read about Dr. Alessandra d'Azzo's NTSAD funded research work, the LOTS Natural History Study at NIH, and the unique partnership with the Quinn Madeline Foundation as they raise funds for the Jacob Sheep, **please revisit last week's issue of "Topic of the Week: Research Review" here**.

FAMILY SUPPORT

The **37th Annual Family Conference** is moving full-steam ahead with 2015 Conference Coordinator, Becky Benson, and Conference VP, Risa Asnen at the helm. They are in the midst of planning sessions to ensure families get everything they need during the three-day weekend. (April 16th to April 19th, 2015 at the Hyatt Regency in Reston, Virginia.)

Currently, the NTSAD Helping Hand Grant Fund needs to be replenished after our successful 2014 Conference in Atlanta. There are two ways to support the Helping Hand Grant Fund - **purchase raffle tickets here** or **make a gift online here**.

NTSAD also welcomes sponsorships for the conference to further enhance the experience for families, children and siblings. Please contact Joan Lawrence at **joan@ntsad.org** for information on the different levels of sponsorship.

MEET CHRIS AND SEAN.

These young men are brothers who are fans of the Maple Leafs and Blue Jays.They enjoy traveling, nature and have their own respective interests in historical biographies Sudoku and word games. Life, however, is challenging due to their struggles with **Late Onset Sandhoff disease**.

While their peers carry on with their lives, Sean (at 30) and Chris (at 28) must watch as the disease restrains them from experiencing true independence. It frustrates them to need help from simple tasks as cutting the meat on their plate or shaving in the morning. The future they envisioned for themselves as teenagers is vastly different. Sean, for example, had to leave college and his dream of being a mechanic due to the disease. Their parents, Debbie and Ralph, are so supportive of them and take every opportunity to travel with Sean and Chris. They've clearly imparted wisdom to their sons as Sean says, "Getting angry isn't going to change anything. We just live each day as it comes."

Please **make a gift** to research today to provide hope to young adults like Sean and Chris!

Their new pursuit - horseback therapy!

UPCOMING NTSAD HAPPENINGS

Mathew Forbes Romer Foundation
An NTSAD South Florida Affiliate
Fourth Annual **Hoops for Hope**
Saturday, October 25, 2014, 1:00pm - 3:00pm
Grandview Prep, 333 Spanish River Blvd. NW, Boca Raton

For a small $10 donation, you can participate in this event. Sign-up **here.**

NTSAD Day of Hope

Every dollar will go to research, so your gift will make a huge impact!

Fourth Annual Day of Hope - Results to Date

Families, their friends and communities have been busy this last month observing the Fourth Annual Day of Hope to rally and raise funds for research! We thank the nearly 1,000 people who have supported these events in some way worldwide! To date **over $35,000** has been raised, bringing us 70% of the way to our goal! **We ask you to consider making a gift here to boost the efforts of our families' events.** If just 100 people give $100 in the next week, we will make it! Stay tuned for the final results in the coming weeks!

THANK YOU.

NTSAD's Vice-President of Finance, Nikki Borman, is stepping down from her position after volunteering her time for over eight years. Her expertise in technology transfer and valuing intellectual property were invaluable as the research landscape that NTSAD funds has expanded.

The NTSAD family extends its deepest gratitude to Nikki and wish her the best in her future endeavors as she focuses on her own company at **Borman & Company** based in the Greater Boston area.

☑ I want to give NTSAD my support today.

Give a gift here.

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STAY CONNECTED