NTSAD Community News

Research, Community, and Collaboration

Supporting families is the center of everything we do...

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Imagine & Believe on Tuesday, November 10th

Need a reason to believe in something good? Join NTSAD on November 10th where we will share advances in research and honor patients and families at NTSAD's annual Imagine & Believe event. Several families will share their stories, experiences, and hopes for the future.

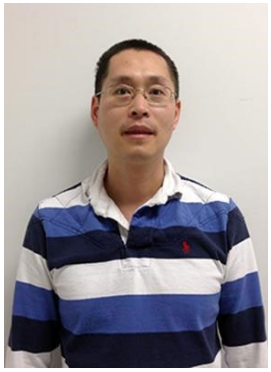
"*Thirteen years ago, I joined NTSAD to drive research. Today, we have at least 10 companies working on drug development with several clinical trials underway and an equal number of academic institutions and companies working on diagnostics, natural history studies, as well as other clinical development tools.* ***We can truly imagine and believe there will be effective treatments for patients and families in the very, near future. This year at Imagine & Believe, we will highlight our powerful community, particularly the families we serve. After all, supporting families is the center of everything we do."*** -- Sue Kahn, NTSAD executive director.

The live event held virtually is accessible from anywhere, making the event larger and more inclusive than ever before. The event will include updates from researchers, partners, and industry members, as well as raise vital funds to support individuals and families affected by Tay-Sachs, Canavan, GM1, and Sandhoff diseases.

To sponsor the event or for more information, please contact Susan Keliher, Director of Development and Communications at skeliher@ntsad.org.

Don't forget to Save the Date for Imagine & Believe!
Tuesday, November 10th, 4:30-5:30 p.m. EST.
Stay tuned for more information at www.ntsad.org.
Free to attend.

Families Fund Research to Identify New GM1 Biomarker



Each year NTSAD families and friends raise money for research by hosting Day of Hope activities. One of several projects funded by their efforts was led by Xuntian Jiang PhD from Washington University School of Medicine. Dr. Jiang identified biomarkers to evaluate the efficacy of gene therapy for patients with GM1 ganglsiodosis.

Dr. Jiang's project specifically identified oligosaccharide biomarkers in patients with GM1 gangliosidosis, a rare, fatal, neurodegenerative genetic disease, to ultimately determine the efficacy of gene therapy. A major challenge for developing treatments for GM1 is the difficulty in evaluating efficacy, which is further complicated by limited patient numbers, heterogeneity in age, severity, and stage of disease progression. Biomarkers provide disease status mileposts to assess the effects of treatment when compared to the natural and unaltered progression of the disease.

In his work, Dr. Jiang identified an oligosaccharide biomarker H3N2b that is significantly elevated in the urine, cerebrospinal fluid (CSF) and plasma from GM1 patients and brains from the GM1 cat model.

After the project was funded by NTSAD, Cindy Erickson, mother to Jorgen, who is 32 years-old and living with GM1, connected with Dr. Jiang seeking to see how she and Jorgen could support his efforts. To help Dr. Jiang isolate enough material to identify the structure of H3N2b, Jorgen donated a sample of his urine.

Thanks to Jorgen, Dr. Jiang and his colleagues were able to synthesize the compound and compare it to urine and plasma samples from the first participant in a Phase 1/2 clinical trial of AAV gene therapy for GM1. They noted a significant reduction of H3N2b and positive response to the treatment, and that H3N2b is a good biomarker for assessment of gene therapy treatment efficacy in GM1 patients.

"NTSAD has supported many important research projects that will lead to treatment and a cure for lysosomal storage diseases including Tay-Sachs, Canavan, GM1, and Sandhoff diseases. I want to acknowledge the support from NTSAD and patient families to my research...The two-year, $80,000 grant from NTSAD allowed me to develop a much-needed tool to facilitate development of treatment for GM1. I would like to give a special thanks to Cindy Erickson, who gave me encouragement and donated the urine sample from her son, Jorgen. Through Jorgen's donation of a biological sample, Jorgen has done so much to help others" **---Dr. Xuntian Jiang.**

Based on the preliminary results obtained from this project, Dr, Jiang submitted an application with the National Institutes of Health (NIH) to continue to use H3N2b as a validated method to assess AAV gene therapy treatment efficacy in the clinical trial over five years, and he recently received approval for this significant grant!

"We are pleased that an NTSAD grant funded by NTSAD families' efforts is being leveraged into larger NIH funding that enables Dr. Jiang's research to be continued for implementation in clinical drug development. We congratulate Dr. Jiang and thank him for bringing his expertise as well as hope to patients and families," said Sue Kahn, executive director of NTSAD.

Dr. Jiang will provide NTSAD with a final report in December which will be shared in a future newsletter.

Axovant Anticipates Approval to Start GM2 Clinical Trial

Axovant anticipates IND clearance of AXO-AAV-GM2, an investigational gene therapy for GM2 gangliosidosis (Tay-Sachs and Sandhoff diseases), from the FDA by the end of 2020. This would mark the start of promising clinical trials as two children who participated in previous expanded access trials continue to demonstrate evidence of disease stabilization. **Read more.**

Aspa Therapeutics Canavan Disease Gene Therapy Webinar

Recently, Aspa Therapeutics, a biotechnology company focused on developing a gene therapy for Canavan disease hosted a webinar for families. Professor Guangping Gao, a pioneer in gene therapy who developed Aspa's investigational gene therapy program, along with a panel of experts provided background about gene therapy, and shared details about Aspa's clinical development program for Canavan disease.

Watch the webinar **here.**

Finding Solace and Strength

As seasons change, children start school, and holidays are observed, many families continue to search for solace. **Carla Steckman, mother of Talia and NTSAD Board member wrote a poignant *Boston Globe* article about grief, acceptance, and reflection during Yom Kippur that** offers valuable perspective for families experiencing grief and anyone struggling during these difficult times. **Read the article.**

Shining a Light on Healthy Sibling Experience

Recently at the **Global Genes Patient Advocacy (Un)Summit, Kyla Marquardt, a rare, healthy sibling shared her story as part of her commitment to honoring her late brother William**. Kyla appeared in Voices of the Siblings, a documentary film produced by Courageous Parents Network (CPN) which was aired as part of three-day film series in partnership with the Disorder Channel. In the documentary, siblings from three families speak poignantly about what it is like to be the brother or sister of a child living with a rare, life-limiting disease.

Following the airing of the film, Kyla and **CPN Founder Blyth Lord**, a mother to two healthy children and daughter Cameron, who passed of infantile Tay-Sachs, participated in a live discussion about their experiences as a healthy sibling, and mother, since the filming of the documentary.

During the live discussion, **Kyla reflected on her younger self, and emphasized the need for healthy siblings like her to receive support as they grieve the loss of shared experiences with a siblings while still honoring them.**

Today, **Kyla Marquardt** is a student at the University of Florida studying chemistry with the goal of become a geneticist. *"My little brother, William, passed away after a long battle with GM1-Gangliosidosis in 2013. I take it upon myself to honor his memory whenever I am able."*

Also, participiating in the documentary film was Jessica Epstein, another healthy sibling and sister of Rachel, who has Canavan.

Currently, NTSAD's Family Services Team is expanding its rare sibling support systems. Interested healthy siblings should reach out to diana@ntsad.org.

CPN is a nonprofit that empowers parents and families caring for children with life-limiting illnesses. Watch the Rare Disease in Children: Voices of the Siblings **here.**

Season of Hope Endures

As autumn rushes in, Day of Hope activities begin to wind down. **We remain grateful to the scores of families and companies who participated in the 10th Anniversary Season of Hope, raising money for research.**

Each year Derrick and Faith Stidham host Eli's Cruise for Cure Car Show to raise funds in honor of their son, Eli who has juvenile Tay-Sachs. To show their support for Eli as well as other children affected by Tay-Sachs, the family includes every child's name on their car's back window.

Employees at PassageBio, a genetics medicine company based in Philadelphia, wore their hearts on their sleeves at a recent team meeting and demonstrated their support of this year's Day of Hope.

"We're proud to support the dedicated work of NTSAD-- allies for patients and their families battling rare diseases. This Day of Hope and every day we celebrate and support advances in gene therapy that directly impact thousands of lives," said Michele Clarke, executive director of patient engagement at Passage Bio.

For the 10th annual Day of Hope, the Romer family held a ping pong tournament in memory of their Superhero Matthew, who passed from Infantile Tay-Sachs in 2003. Day of Hope unites all families as we honor and remember those we love.

Let's hear it for Team M&M for their successful Day of Hope event! Congratulations the Ronaldson family and their community of supporters for raising more than $26,000 this year alone for research via a virtual walk/run.

NTSAD's Sue Kahn and Diana Pagonis joined Team M&M participating in the virtual event from New England and in support of Ronaldson Family as they raise awareness and research funds for Juvenile Sandhoff disease.

NTSAD leads the worldwide fight to treat and cure Tay-Sachs, Canavan, GM1, and Sandhoff diseases by driving research, forging collaboration, and fostering community. Supporting families is the center of everything we do.

Donate

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