NTSAD Community News

Research, Collaboration, and Community

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

March 2021

In this Issue

Taysha Begins Enrollment for GM2 Clinical Trial

Taysha Gene Therapies begins enrollment for the company's TSHA-101 gene therapy clinical trial for infantile GM2 gangliosidosis (Tay-Sachs and Sandhoff diseases) at Queens University in Kingston, Ontario, Canada.

Learn more about the clinical trial.

TAYSHA GENE THERAPIES

Passage Bio's GM1 Clinical Trial Receives Fast Track Designation from FDA

The U.S. Food and Drug Administration (FDA) has granted Fast Track Designation to three of Passage Bio's lead investigational gene therapies, including PBGM01 for the treatment of GM1 gangliosidosis, and the company is actively recruiting at one site to date.

Read press release.

For more information about the study.

PassageBio

Tay-Sachs Gene Therapy Uses Innovative Delivery Approach

NTSAD is a proud partner and investor in the translational research of Dr. Miguel Sena-Esteves, PhD and Heather Gray-Edwards, DVM, PhD, both of UMass Medical School, along

with Douglas Martin, PhD at Auburn University College of Veterinary Medicine and other investigators in the Tay-Sachs Gene Therapy Consortium, that led to gene therapy clinical trials for infantile GM2 (Tay-Sachs and Sandhoff diseases). In parallel, Drs. Sena-Esteves and Martin also were the lead investigators for GM1 gene therapy. **The research findings were licensed and further developed by Sio Gene Therapies, formerly Axovant. The first patient in the company's clinical trial for AXO-AAV-GM2 was dosed using an innovative neurosurgical technique to overcome the blood-brain barrier.**

Oguz Cataltepe, MD, professor of neurological surgery, whose pediatric neurosurgery team delivered the therapy, found the new technique enables better drug delivery to brain tissue, when compared to a drug being injected into cerebral spinal fluid spaces using lumbar puncture or intraventricular delivery techniques. This new neurosurgical technique has only been used a few times in other clinical trials including for Parkinson's and brain tumors.

Read more about the innovative neurological technique.

Recently, Dr. Sena-Esteves shared his expertise and experience in the development of the first in-human clinical trial of AAV gene therapy for Tay-Sachs Disease at Quinnipiac School of Medicine's Rare Disease Day event.

Watch Dr. Sena-Esteves' Address.

Late Onset FDA Listening Session Summary

In January, members of the Late Onset Tay-Sachs and Sandhoff (LOTSS) community participated in a listening session with the U.S. Food and Drug Administration (FDA), so regulators could better understand the impact of heterogenous disease on the lives of individuals and their families. Several affected individuals and two family members shared their personal stories, including symptoms, years to diagnosis, and their collective hope to slow the progression of the disease and maintain their independence. NTSAD thanks members of the LOTSS community for their advocacy and tireless efforts toward finding effective treatments. We are pleased to share a summary of the presentation to further promote their efforts.

The listening session was dedicated to a much beloved member of the NTSAD LOTSS community, Scott Hunger, who passed away on December 21, 2020.

We plan to organize future FDA Listening Sessions about GM1 gangliosidosis, Canavan, and Infantile and Juvenile GM2.

Read summary here.

Register Now for NTSAD's 43rd Annual Family Conference

Join nearly 400 families and members of the NTSAD community for the 43rd Annual Family Conference on April 22-25, 2021. The event is held virtually and is free to attend. The conference is open to any family coping with a Tay-Sachs, Canavan, GM1 Gangliosidosis, or Sandhoff diagnosis, as well as members of industry, researchers, clinicians, and rare allies. All are welcome!

View the Conference Schedule.

For families living in the U.S., please register by March 26 to receive a complimentary conference in a box. All other Community members please register by April 8.

Register Now

When registering, please note some sessions are specifically created for and limited to rare individuals, such as members of the Late Onset community, or specific rare family members i.e. parents, siblings, or grandparents and extended family members. Many sessions are open to all attendees and are indicated as such.

The Research Session is open to all and will be held on Friday, April 23 from 12-4 p.m. EDT. Friday's Session focuses on the latest advances in research and clinical trials and includes breakout sessions by disease.

Throughout the four-day conference, sessions are held via Zoom in either a Webinar or meeting format and hosted in English during Eastern Daylight Time. Prior to each session, Zoom links are sent via email and posted on NTSAD's website (**www.ntsad.org**) and on the Conference's Crowd Compass app.

No matter where we are in the world, during the conference we come together--

Heart to Heart, Home to Home!

NTSAD Gratefully Acknowledges 2021 Annual Family Conference Sponsors*

COURAGE

Sanofi Genzyme

HOPE

Recursion

Laura and Simeon Schindelman

Taysha Gene Therapies

INSPIRATION

Aspa Therapeutics

Drive FORE Dylan

Lysogene

Mathew Forbes Romer Foundation

Passage Bio

The Quinn Madeleine Foundation

Sio Gene Therapies

DETERMINATION

Amicus Therapeutics

Azafaros B.V.

Cameron & Hayden Lord Foundation

Gain Therapeutics

Idorsia Pharmaceuticals, Ltd

COMMUNITY

B Brave Foundation

Bonnie and Barry Davis in loving memory of Adam Davis

IntraBio

***Sponsors as of publication date.*
Please forgive any omissions or errors.

Choosing Resiliency in the Face of Adversity

Watch Sheryl Sandberg, Facebook COO, founder of Lean In, and co-author of Option B: Facing Adversity, Building Resilience, and Finding Joy, and Blyth Lord, Founder of Courageous Parents Network, in a conversation about the challenges they both faced and what they learned following unexpected, life-altering situations. Sheryl and Blyth along with Becky Benson, NTSAD's Family Services and Conference Coordinator and mom to Miss Elliott, discussed anticipatory grief, the importance of being mindful of things that bring you joy even during hard times, and leveraging post-traumatic growth.

More than 200 people participated in this poignant and inspiring conversation filled with personal stories and examples of how to build resiliency and support others through difficult experiences.

Watch the conversation.

The Flysjö Family Discuss and Film Their Clinical Trial Experience

It's been almost a year since the Flysjö family rushed from their home with their three small children as countries were shutting down borders due to the COIVD pandemic, so Hampus, Isabella, and Julia could participate in GM1 gangliosidosis gene therapy clinical trial at NIH. Recently, their parents Jessica and Niclas spoke with Cynthia J. Tifft, M.D., Ph.D., Deputy Clinical Director of the Office of the Clinical Director at the National Human Genome Research Institute (NHGRI), about their experience and their partnership as part of NIH's Rare Disease Day presentations.

Throughout their conversation, **Jessica and Niclas shared their journey to finding a diagnosis, their family's perilous journey from Sweden, and the relief they felt after their three children received the gene therapy.**

Read a synopsis about the event.

Watch and listen the conversation with the Flysjö Family.

(Please note to watch the conversation begin at the 1 hour and 30 minute mark.)

For more on the Flysjö Family's experience, watch The B Brave Foundation's docuseries and view 12 mini-episodes filmed by the family in real time, while they were living at NIH and the children received treatments.

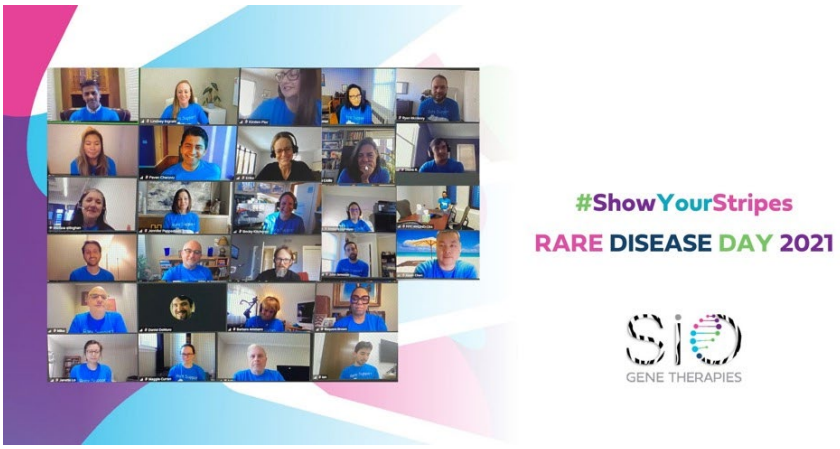
In the final episode, parents Jessica Lindqvist and Niclas Flysjö discuss their children finally being approved to undergo gene therapy for Late-Infantile GM1 and their feelings surrounding this critical step.

Watch Episode 12.

The B Brave Foundation's Project Rare Docuseries is a collection of films featuring families, caregivers and healthcare professionals wrestling with the complexities of grief, clinical trials, and caring for loved ones with incurable diseases.

We All Care for Rare

On February 28, people around the world shared their rare stories to advocate and raise awareness for rare diseases. Thank you to families and rare allies for your participation and support.



Sio Gene Therapies showed their support for rare by connecting with a family directly impacted by Tay-Sachs disease who shared their story, while Sio employees sported their NTSAD's Rare Support Bear t-shirts.

Rare Bear t-shirts are still available! **Purchase a shirt now.**

Check out more Rare Disease Day stories, events, and news.

Motherhood and Parenting a Rare Child

Becky Benson, NTSAD's Conference and Family Services Coordinator and mom to Miss Elliott, is a published author. In a recent essay published in Months to Years Becky shares that before receiving her daughter's Tay-Sachs diagnosis, she encountered feelings of inadequacy in her ability to connect with her new baby, due to the nagging sensation that something was very wrong. In this poignant essay, Becky describes her perceived insecurities and preliminary feelings of failure as a mother before ultimately learning of the diagnosis that would change their lives forever.

Read Becky's article.

Family Ancestry and Carrier Screening

Anyone can be a carrier of Tay-Sachs, Canavan, GM1, and Sandhoff diseases. When both parents are carriers, each child has a 25% of having the disease. The carrier rate for the general population for GM1 and Tay-Sachs diseases is 1/250. **Some evidence suggests people of Irish / British Isle descent have an increased risk over the general population between 1/50 to 1/150. French Canadians, Louisiana Cajuns, and Ashkenazi Jews are all considered high risk with a carrier rate of 1/27.**

For Canavan disease the carrier rate for the general population is 1/300. Ashkenazi Jews are at high risk with a carrier rate of 1/55. For Sandhoff disease the carrier rate is approximately 1/600 in the general population, and it is not yet clear whether Sandhoff disease is more common in any particular population, but it may have a higher carrier rate in several, somewhat isolated populations.

Begin a conversation about the importance of carrier screening with your family and in your community.

Read more about carrier screening.

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

STAFF
Sue Kahn, Executive Director
Becky Benson, Family Services and Conference Coordinator
Sydnie Dimond, Development and Communications Associate
Susan Keliher, Director of Development and Communications
Diana Pangonis, Director of Family Services

NTSAD
2001 Beacon Street
Suite 204
Boston, MA 02135

info@ntsad.org
www.NTSAD.org