NTSAD Community News

Research, Collaboration, and Community

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

April

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Dear NTSAD Community,

It is with great anticipation and excitement that I share that our first in-person Imagine & Believe event to be held in three years is Thursday, November 10 in Boston where we will commemorate NTSAD's 65th Anniversary! This year we are delighted to honor research pioneer Guangping Gao, PhD., whose astounding professional legacy is resurrecting gene therapy and potential treatments for many current untreatable human diseases, including Canavan disease. Dr. Gao's Canavan research has been licensed by Aspa Therapeutics for the company's current clinical trial.

Earlier this month, Research Director Valerie Greger, PhD, Director of Family Services Diana Pangonis, and I took a trip to the University of Massachusetts Chan Medical School (UMASS) and had the pleasure of visiting with Dr. Gao, along with two other extraordinary, rare disease researchers Dr. Heather Gray-Edwards PhD and Miguel Sena-Esteves PhD.

Last fall, NTSAD awarded a grant to Dr. Gray-Edwards to sustain a flock of Jacob sheep who have a naturally occurring form of Tay-Sachs disease similar to that in humans with the late onset form of the disease. Keeping the flock alive enables the next generation of researchers to test treatments. During our visit to UMASS, we had the opportunity to observe the flock of Tay-Sachs sheep participating in the gene therapy research study and could not resist the urge to snap some photos. Visiting with these esteemed researchers, touring the site, and meeting the sheep and lambs was a very special experience! During the last 12 years, NTSAD has invested more than $400,000 toward research involving the Tay-Sachs disease (TSD) sheep model.

Finally, I would be remiss if I did not thank all of our sponsors and supporters of NTSAD's 44th Annual Family Conference in Denver this July. Your sponsorship along with Helping Hand grants makes the conference affordable for families to attend.

Chag Pesach Sameach and Happy Easter to all who celebrate!

Kathy

Kathleen M. Flynn
Chief Executive Officer

Learn more about the sheep.

44th Annual Family Conference July 7-10, 2022

If you or a loved one are coping with a diagnosis of Canavan, GM1, Sandhoff or Tay-Sachs disease, or you are a caregiver, researcher, industry member, or clinician, we invite you to join us at the conference on July 7 to 10 in Denver, Colorado.

"The conference has been a wonderful resource for our family. The content is always excellent, and what we've found equally valuable are the friendships we've made with other families. Parenting an affected child can be isolating, and we can't overstate how nice it is to be able to spend time with other parents who get it." -Ashlee Suran, Mom to Cameron, who has Canavan disease.

Don't miss the conference, register today.

Helping Hand grants are available for families in need of financial assistance.

Read the COVID guidelines for the conference.

Thank you again to the 2022 Annual Family Conference Sponsors who make it possible for NTSAD to provide families with connection, community, and support.

Learn how you can support the conference.

PRESENTING

The Doyle Foundation

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Courage

Laura and Simeon Schindelman

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EveryLife Foundation

The Cameron & Hayden Lord Foundation*

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In Memory of
Dylan Manning and Stewart Altman

Mathew Forbes Romer Foundation

Passage Bio

Joyce and Edward Ratner

Inspiration

Judy Kaplan

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Connection

Staci Kallish* and Chris Beer

Propel Careers*

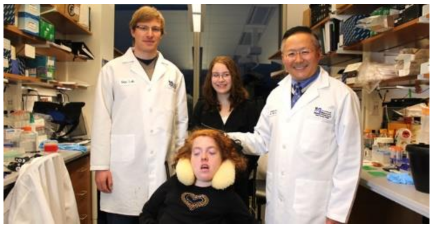
*NTSAD Board Member

Imagine & Believe 2022 Honors Research Pioneer Guangping Gao

On Thursday, November 10, 2022, the NTSAD Community will gather at the Royal Sonesta in Boston for the Imagine & Believe event to commemorate NTSAD's 65th Anniversary and raise funds for programs and services. At the event NTSAD will honor research pioneer Guangping Gao PhD for revitalizing gene therapy and potential treatments for rare diseases, including his research toward treatments for Canavan disease.

Dr. Gao is the co-director, Li Weibo Institute for Rare Diseases Research, Director, Horae Gene Therapy Center and Viral Vector Core, Professor of Microbiology and Physiological Systems, Penelope Booth Rockwell Professor in Biomedical Research, University of Massachusetts Chan Medical School. Dr. Gao started studying Canavan disease in 1989 and discovered the gene and its mutations in 1993. He discovered the vector (AAV9) for efficient delivering the therapeutic gene to the brain via intravenous injection in 2004 and developed an optimized effective Canavan gene therapy in 2017. His work was licensed by Aspa Therapeutics where clinical trials have begun.

His dedication and compassion are unparalleled as many families can attest. NTSAD remains grateful to Dr. Gao for his many contributions benefitting the NTSAD Community–from establishing the Gao Lab at the Horae Gene Therapy Center at UMASS, to mentoring the next generation of researchers, and serving as a trusted expert for families.



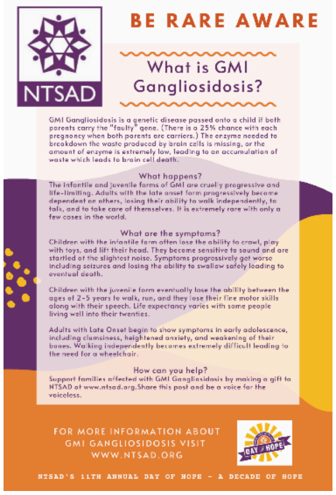
Furthermore, Dr. Gao is prolific in the number of biotech patents he holds, which total more than 130 with additional patents pending, as well as many research papers and publications. He also is the former President of the American Society of Gene and Cell Therapy and co-founded Voyager Therapeutics. Recently *BioSpace*, a digital magazine, published an article about Dr. Gao highlighting his career and two life-changing experiences, including working on a farm, and living in a barn in China as a teen, and meeting a Canavan family 15 years ago (Sherri Sigel and her two daughters Rachel Epstein, who has Canavan, and Jessica who later worked in Dr. Gao's lab). **Read the article.**

Please save the date and join us on November 10 in honoring Dr. Gao and commemorating NTSAD's 65th Anniversary at the Imagine & Believe event.

International GM1 Gangliosidosis Awareness Day: May 23, 2022

Get involved in the first International GM1 Gangliosidosis Awareness Day:

- If you care for an affected loved one, share your story and a picture on social media.
- **Add the GM1 Awareness Day frame to your Facebook profile picture.**
- Share NTSAD's GM1 Be Rare Aware Infographic on social media.
- Follow NTSAD on **Facebook**, **Linkedin**, **Twitter**, and **Instagram**, and encourage others to do so too!
- **Donate to support affected families.**



NTSAD

BE RARE AWARE

What is GMI Gangliosidosis?

GMI Gangliosidosis is a genetic disease passed onto a child if both parents carry the "faulty" gene. (There is a 25% chance with each pregnancy when both parents are carriers.) The enzyme needed to breakdown the waste produced by brain cells is missing, or the amount of enzyme is extremely low, leading to an accumulation of waste which leads to brain cell death.

What happens?

The infantile and juvenile forms of GMI are cruelly progressive and life-limiting. Adults with the late onset form progressively become dependent on others, losing their ability to walk independently, to talk, and to take care of themselves. It is extremely rare with only a few cases in the world.

What are the symptoms?

Children with the infantile form often lose the ability to crawl, play with toys, and lift their head. They become sensitive to sound and are startled at the slightest noise. Symptoms progressively get worse including seizures and losing the ability to swallow safely leading to eventual death.

Children with the juvenile form eventually lose the ability between the ages of 2-5 years to walk, run, and they lose their fine motor skills along with their speech. Life expectancy varies with some people living well into their twenties.

Adults with Late Onset begin to show symptoms in early adolescence, including clumsiness, heightened anxiety, and weakening of their bones. Walking independently becomes extremely difficult leading to the need for a wheelchair.

How can you help?

Support families affected with GMI Gangliosidosis by making a gift to NTSAD at www.ntsad.org.Share this post and be a voice for the voiceless.

FOR MORE INFORMATION ABOUT GMI GANGLIOSIDOSIS VISIT WWW.NTSAD.ORG

DAY of HOPE

NTSAD'S 11TH ANNUAL DAY OF HOPE - A DECADE OF HOPE

(Right click to save the graphic)

Granted: A Wish Story

Join us in supporting NTSAD Dad and filmmaker Dan Redfield, whose very personal film, *Granted: A Wish Story*, made in honor of his daughter Ava Rose, is now available on Amazon Prime!

Ava Rose had Tay-Sachs disease and passed away in November of 2021. This inspiring and poignant film captures the adventures Ava shared with her parents and little sister Reagan in their home state of Alaska.

Dan shares, *"It's been nearly two years since we started on our Make-A-Wish journey with Ava. To be able to release this to the public is surreal and final. This project was an absolute undertaking and for many months all consuming, physically, and emotionally. My goal was to give Reagan the things I don't have much of, captured memories of her sister. Since* *she's too young. I hope this film serves as proof of her time outside with her big sister. I hope she will watch it with smiles as she gets older."*

Watch Granted: A Wish Story and leave a review on Amazon!

In addition to the film Dan and his wife Kristen have a foundation to help other children like Ava and their families to capture memories and have an adventure of their own.

Read more about the Foundation.

Watch Indie Alaska's video on the Foundation.

Remembering Stewart Altman

The NTSAD Community recently lost a beloved member. Late Onset Tay-Sachs and Sandhoff disease advocate and former NTSAD Board member Stewart Altman. This past February, Stew and Lorrie Altman shared with *Patient Worthy* how they built a beautiful life while navigating the challenges presented by a Late Onset diagnosis. In honor of Stew, here are parts one and two of their interview.

Part 1 and **Part 2**

NTSAD's Director of Family Services Diana Pangonis shared memories of Stew. *"I will miss my friend, dearly. For those who knew Stew, you knew his humor. It makes me smile just thinking about him. One thing is most certainly clear - Late Onset Tay-Sachs did not define Stew in any way, nor did any of his other health challenges. His beautiful spirit and tenacity, the joy you saw in his eyes and his smile, and his love for his community shined through it all. He will be missed. Tremendously. Undeniably. I'm so glad I have his 'what's doing' phone calls and all the years I've known him in my heart. Sending love to Lorrie, his sons, and their families, Rich, and sweet Chipper."*

The NTSAD Community will keep Stew in our hearts and try our best to #BeLikeStew.

Auction, Auction!

NTSAD is seeking donations of items and experiences for a silent auction at Imagine & Believe as well as volunteers who can help solicit and secure them. Items such as bottles of wine; gift certificates for dining at restaurants and chef's tables; airline vouchers; travel opportunities including vacation stays and getaways; theater (Broadway!), concert, and sporting event tickets and memorabilia; unique experiences; jewelry; and spa packages will help us to raise money for programs and services for families. Event guests live in Boston and out-of-town, so we are seeking physical items that may be easily shipped as well as gift cards and opportunities. If you are interested in donating, or in volunteering for NTSAD auction committee, please contact Development and Communications Associate Sydnie Dimond at **sdimond@ntsad.org**

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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