NTSAD Community News

Research, Collaboration, and Community

Supporting families is the center of everything we do…

June

June 2023

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45th Annual Family Conference

More than 300 people participated in the 45th Annual Family Conference in Reston, Virginia, from June 1 to 4. Nearly 80 Rare families were represented, including 14 attending for the first time. We had a record 28 children affected by Tay-Sachs, Canavan, GM1, and Sandhoff diseases, who were provided professional and loving care by nurses and volunteers who worked tirelessly in Camp Snuggle and Camp Active. In addition, we brought researchers, clinicians, and patient advocates together at an industry roundtable, scientific meeting, and during a full day of research sessions where they presented their research and met with families throughout the four-day event.

This year's Conference also included a record number of families from Latin America, including Argentina, Chile, and Mexico. They made vital connections with one another and participated in family and research sessions, with the help of three Spanish-speaking professionals from the National Institutes of Health who provided free translation services. Luis Fernandez and Alejandra Saipert, parents of Giuli and Facu and founders of Cutasa, a patient advocacy

group, will share information from the Conference with more than 50 families living in Argentina and coping with a diagnosis of Tay-Sachs or Sandhoff disease.

A BIG THANK YOU to all the sponsors, speakers, volunteers, and nurses, who helped to make the Conference meaningful for all. A special thank you to Holly Stringer and Karen Horton, R.N. for making the camps a memorable and safe experience for the children and a much-needed respite for parents. Thanks to photographers Sarah Mattingly and Jaime Carpenter for capturing special moments by taking scores of beautiful family portraits. We are grateful to Kate Priest, LCSW, for offering professional support and comfort to our families again this year.

SAVE THE DATE: The 46th Annual Family Conference will take place from April 11 to 14, 2024, in Chicago, Illinois, at the Hilton Oak Brooks Hills Resort. We look forward to seeing you!

Annual Family Conference: Thank You Sponsors!

The generosity of our 2023 Conference Sponsors makes it possible for Rare children, adults, and their families to experience four days of connection, community, and support.

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Annual Family Conference Research Day

At every Annual Family Conference, Friday is devoted to Research where researchers and members of industry members share the latest updates on their research and drug development efforts for Tay-Sachs, Canavan, GM1 gangliosidosis, and Sandhoff diseases with families. During disease-specific breakout sessions, they gather information and ask presenters questions about the studies. Below is a summary of this year's presentations.

Research sessions were not recorded due to proprietary information.

Infantile/Juvenile GM1 & GM2 Session

Doug Martin, PhD, from Auburn University and **Florian Eichler, MD, from Massachusetts General Brigham Hospital** moderated the robust infantile/juvenile GM1 and GM2 breakout session, where six researchers and members of industry developing treatments presented the latest findings on their work.

- **Cynthia Tifft MD, PhD, from the National Institutes of Health (NIH)** discussed the first-in-human Phase 1-2 gene therapy clinical trial for GM1.
- **Kristen Skvorak, PhD, from Codexis** shared data from the company's preclinical research focused on engineering beta-galactosidase enzyme variants for GM1 gene therapy or enzyme replacement therapy.
- **Christian Freitag, MD, from Azafaros** discussed the company's ongoing studies of the oral drug AZ-3102 for GM2. **Read about Azafaros' PRONTO study.** **Read about Azafaros' RAINBOW study.**

- **Ryo Higurashi from JCR Pharmaceuticals** presented findings from the development of a brain-penetrating enzyme replacement therapy for treating Tay-Sachs disease.

- **Isabela Batsu, MD, from Sanofi** discussed findings from the clinical development of Venglustat as a GCS inhibitor for treating GM2 and related diseases.

- **Terence Flotte, MD, from the University of Massachusetts Chan Medical School** reported on the clinical dose-response relationship and safety profile of gene therapy for GM1 and GM2.

- **NTSAD Board Member Gerry Cox, MD, PhD**, discussed NTSAD's pending request to host a GM2 Externally Led Patient-Focused Drug Development meeting with the Food and Drug Administration (FDA), to bring to the forefront the concerns, priorities, and urgency of families with affected children to the FDA and the agency's decision makers regarding treatments.

Canavan Session

Orren Alperstein, President of the Canavan Foundation, moderated the Canavan breakout session, which included updates on ongoing clinical trials with encouraging preliminary results as well as a summary of an FDA meeting and two presentations on NTSAD's Research Initiative's newly funded grants.

- **Olga Flamini MD, PhD, from Myrtelle** presented ongoing results from the company's Canavan disease gene therapy trial.

- **Genevieve Laforet MD, PhD, from Aspa Therapeutics** provided an update including promising data from the company's ongoing gene therapy Phase 1-2 clinical trial.

- **NTSAD Research Associate Caroline Aragón, MS, CGC,** presented a summary of key themes from NTSAD's recent Canavan Listening session with the FDA held this spring.

- **Amanda Nagy, MD, from Massachusetts General Brigham Hospital and Dominic Gessler, MD, from University of Massachusetts Chan Medical School,** recent recipients of NTSAD's Research Initiative grant awards, shared an overview of their upcoming respective research projects. Dr. Nagy's project focuses on reviewing MRI data to understand changes in the brain in Canavan disease. Dr. Gessler will develop an artificial intelligence system to learn disease progression and guide therapy decision-making.

Late Onset Tay-Sachs and Sandhoff (LOTSS) Session

Staci Kallish, DO, from Children's Hospital of Pennsylvania and Immediate Past President of NTSAD's Board of Directors, moderated the Late Onset Tay-Sachs and Sandhoff (LOTSS) breakout session which highlighted exciting progress towards new therapies and a presentation on one of NTSAD's Research Initiative's newly funded grants.

- **Julie Kissell, a PhD candidate from the University of Wisconsin,** and a 2023 NTSAD Research Initiative grant recipient, presented an overview of her project to develop a disease-specific clinical rating scale for late-onset GM2 gangliosidoses.

- **Taylor Fields from Intrabio** updated the community on the clinical trial progress for IB1001, N-acetyl-L-leucine.

- **Heather Gray-Edwards, DVM, PhD, from University of Massachusetts Chan Medical School** and Co-Chair of NTSAD's Scientific Advisory Council reported on the ongoing clinical trial developing AAV gene therapy for LOTSS.

- **Camilo Toro, MD, from the National Institutes of Health** and on behalf of Sanofi presented progress on the clinical development of Venglustat in GM2 and related diseases.

45th NTSAD Annual Family Conference Awards

Each year, NTSAD recognizes an individual or group who are not directly affected by a rare disease, and yet have dedicated countless hours of service and support to our community. This year, **NTSAD awarded Mimi Blitzer, PhD, and Erin Strovel, PhD, the Above & Beyond Award for their efforts that led to the completion of NTSAD's Tay-Sachs Testing Quality Control Program following the program's founder, Michael Kaback, MD.** Dr. Blitzer and Dr. Strovel's important work ensures laboratories give accurate genetic carrier screening results to carriers of Tay-Sachs disease and provide support, information, and options, so people may safely plan their families.

Every year at NTSAD's Annual Family Conference we recognize someone who has been affected by a rare disease and selflessly supports other rare families through personal service. This year, **Holly Stringer, Mom of Brennan, was recognized with NTSAD's Power of One Award for creating the Conference's Camp Activities Program for affected kids.** Each year, Holly leads a team of volunteers and nurses in an extraordinary effort to not only care for the kids, but to provide them with a camp-like experience that includes playing games, creating arts and crafts, and most of all making new friends and having fun. For older affected children, this is often the only time they receive a camp experience. Thank you, Holly, for all you do for rare kids and their families!

Azafaros' GM2 Clinical Trial Update

Azafaros recently announced that the first patient has been enrolled in the company's Phase 2 RAINBOW study for GM2 (Tay-Sachs and Sandhoff diseases). The clinical trial is being conducted in Brazil and the U.S. and will evaluate the safety, tolerability, pharmacokinetics, and pharmacodynamics across two doses of its lead asset, AZ-3102, in patients with GM2 gangliosidosis.

Learn more.

Thank You for Your Board Service

This June, two members of NTSAD Community, Ruth Feldman and Amy Speak, were honored for their dedication and service as members of the Board of Directors.

Ruth Feldman, whose brother Aaron had infantile Tay-Sachs, served for a decade on the Board with a commitment to healthy siblings both children and adults. When healthy siblings ranging from young teens to adults came together several years ago to support and remember loved ones, Ruth shared she had waited decades to connect with other siblings, which she continues to do including at the most recent Annual Family Conference.

During Ruth's tenure on the Board, she served as Secretary for three years and as a member of the Board Development Committee. Ruth also played a vital role in NTSAD's 2019 Strategic Plan process and as a member, and later the Chair of the Development (Fundraising) Committee. She was recognized by the Board's leadership for providing valuable guidance on best practices and Board governance throughout her time on the Board. Ruth will continue serving on the Development Committee. Ruth, we thank you!

Amy Speak served on the Board for six years and as a valued member of the Communications Committee. The Chair of the Committee and fellow Board member Jonathan Katz paid tribute to Amy at her last Board meeting, saying:

"*Author Zig Ziglar once said,* ***'You never know when a moment and a few sincere words can have an impact on a life.' This quote perfectly captures the essence of Amy's contributions. Amy's dedication, passion, and exceptional communication skills have positively impacted our organization and community.***

Amy understands the profound power of communication in effecting positive change. She recognizes that thoughtful and purposeful communication can help connect people in meaningful ways and motivate people to take action. Amy understands that being an effective communicator is not about dominating conversations, but about being an active listener. Amy is an outstanding listener. In part, this is what enables her to provide guidance that makes a real difference and helps amplify voices to support meaningful advocacy.

Amy's tenure as a board member is marked by a commitment to our organization, and genuine compassion for the community we serve. As we reflect upon her contributions, let us remember the impact that can be achieved through sincere words and genuine connection. Amy's service to the board provides an important reminder of the transformative power of communication, inspiring us all to listen, empathize, and communicate with purpose."

Jonathan, we couldn't have said it better. Thank you, Amy for your service. Amy will continue to serve on the Communications Committee.

Move a Mile for Hope in Style!

Join the NTSAD community in raising awareness and funds this year's Day of Hope, September 9th! Our goal is to raise a minimum of $50,000 for research. Will you join us?

You can participate and Move a Mile for Hope! Gather friends and family and colleagues to join you for a walk, roll, or stroll, around your neighborhood, hosting a dance party, or even a scenic drive.

So you can represent the Rare NTSAD Community in style when you Move a Mile for Hope or whenever you wear your sneakers, **NTSAD has created special, purple, Move a Mile for Hope shoelaces!** Contact NTSAD's Development and Communications Manager Sydnie Dimond at **sdimond@ntsad.org** to request purple laces for you, your family, or co-workers.

Get involved:

- **Mark your calendar now to Move a Mile for Hope on September 9th!** Remember to post photos of your new laces and raise awareness online. Tag @NTSAD!
- Fundraise for Research via Facebook fundraiser, an NTSAD donation link, or by selling laces! (Contact **Sydnie** for assistance!)
- Host an event that is entirely your own! (Contact **Sydnie** for ideas and support!)

NTSAD Research Initiative: 2023 Research Grant Recipients

On Friday, June 2, at NTSAD's 45th Annual Family Conference during Research Day, Research Director Valerie Greger, PhD, announced NTSAD's Research Initiative's 2023-2024 grant awardees whose work meets our goal to accelerate research leading to effective treatments for Tay-Sachs, Canavan, GM1, and Sandhoff diseases. **This year, NTSAD funded clinical research projects that focus on tools that support future studies and trials, including the development of severity scales for disease staging, translational biomarkers with clinical utility, and measurable and meaningful efficacy endpoints for new therapeutic approaches.**

The 2023-2024 Research Grant Projects and Principal Investigators are:

- Characterization of Progressive Neuroimaging and Pathologic Changes in Canavan Disease (Amanda Nagy, MD; and Florian Eichler, MD)
- Non-Invasive MRI-based Therapeutic Outcome Prediction Modeling Using Machine Learning (Dominic Gessler, MD, and Guangping Gao, PhD)
- Development of a Disease-Specific Clinical Rating Scale for the Late-Onset GM2 Gangliosidoses (Jennifer Kwon, MD, and PhD candidate Julie Kissell)

NTSAD is grateful to the **Canavan Foundation** for its partnership and support of these grants.

NTSAD's Research Initiative began in 2002 and has awarded 69 grants to date, providing more than $4.1 million in funding. During this funding cycle, the Research Initiative received 15 pre-applications, and four investigators were invited to submit full proposals, each of which underwent a rigorous review by NTSAD's scientific advisors and outside experts. Thank you to our grantees, applicants, research and review committees, partners, donors, and, of course, families for inspiring these projects. Together, we are advancing research.

Learn more about NTSAD's Research Initiative and grant opportunities.

Honoring Rare Dads on Father's Day

On Father's Day, NTSAD recognizes the rare love of all the dads in our rare community. Whether you are actively caring for or grieving a child or grandchild, we see you and appreciate all you do.

If you know a father who is grieving, say his child's name. Acknowledge his pain and the enduring love he has for his child. Today, we celebrate the special bond between a father and his child. We also send love to all who are grieving the loss of their father.

International Newborn Screening Awareness Day

The first ever International Newborn Screening Awareness Day is June 28, 2023! *Rare Revolution Magazine's* most recent issue explores newborn screening programs in America, Europe, and Australia, highlighting the vital role they play in society.

Read the issue.

Texas Governor Signs Landmark Bill Paving the Way for Newborn Screening

On June 13, Texas Governor Greg Abbott affirmed the state's status as a leader in newborn screening by signing House Bill 2478 into law. The law, referred to as RUSP (Recommended Uniform Screening Panel) alignment legislation, implements a three-year timeline in which the screening must begin for new conditions added to the federal RUSP, and ensures that the Texas Department of State Health Services shall provide an annual report to state leadership that outlines the department's capacity to implement additional nationally recommended newborn screening tests, including the ability to add conditions within two years of addition to the RUSP.

Every year, hundreds of thousands of babies born in Texas are screened immediately after birth to detect life-threatening conditions that require urgent medical intervention. More than 1,200 newborns receive life-saving diagnoses through newborn screening, but unfortunately, Texas currently does not screen newborns for several recommended conditions. This delayed implementation creates disparities in clinical care and leaves hundreds of babies undetected and without timely treatment. *"Early diagnosis and intervention through newborn screening save lives,*" explained Tiffany House, a Texas advocate and president of the Acid Maltase Deficiency Association. "*Even a few days difference in the initiation of treatment can have a profound impact on the path that these children's lives will take."*

Texas is the 11th state to expand Newborn Screening through legislation in recent years.

Advancing newborn screening for Tay-Sachs, Canavan, GM1 and Sandhoff diseases remains a priority for NTSAD and our Newborn Screening Consortium.

To learn more about the legislation and how to support newborn screening, visit **RareScreening.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

STAFF
Kathleen M. Flynn, CEO
Caroline Aragón, MS, CGC, Research Associate
Becky Benson, Family Services Manager
Sydnie Dimond, Development and Communications Manager
Valerie Greger, Director of Research
Diana Jussila, (formerly Pangonis) Director of Family Services
Susan Keliher, Director of Development and Communications

NTSAD
2001 Beacon Street
Suite 204
Boston, MA 02135

info@ntsad.org
www.NTSAD.org