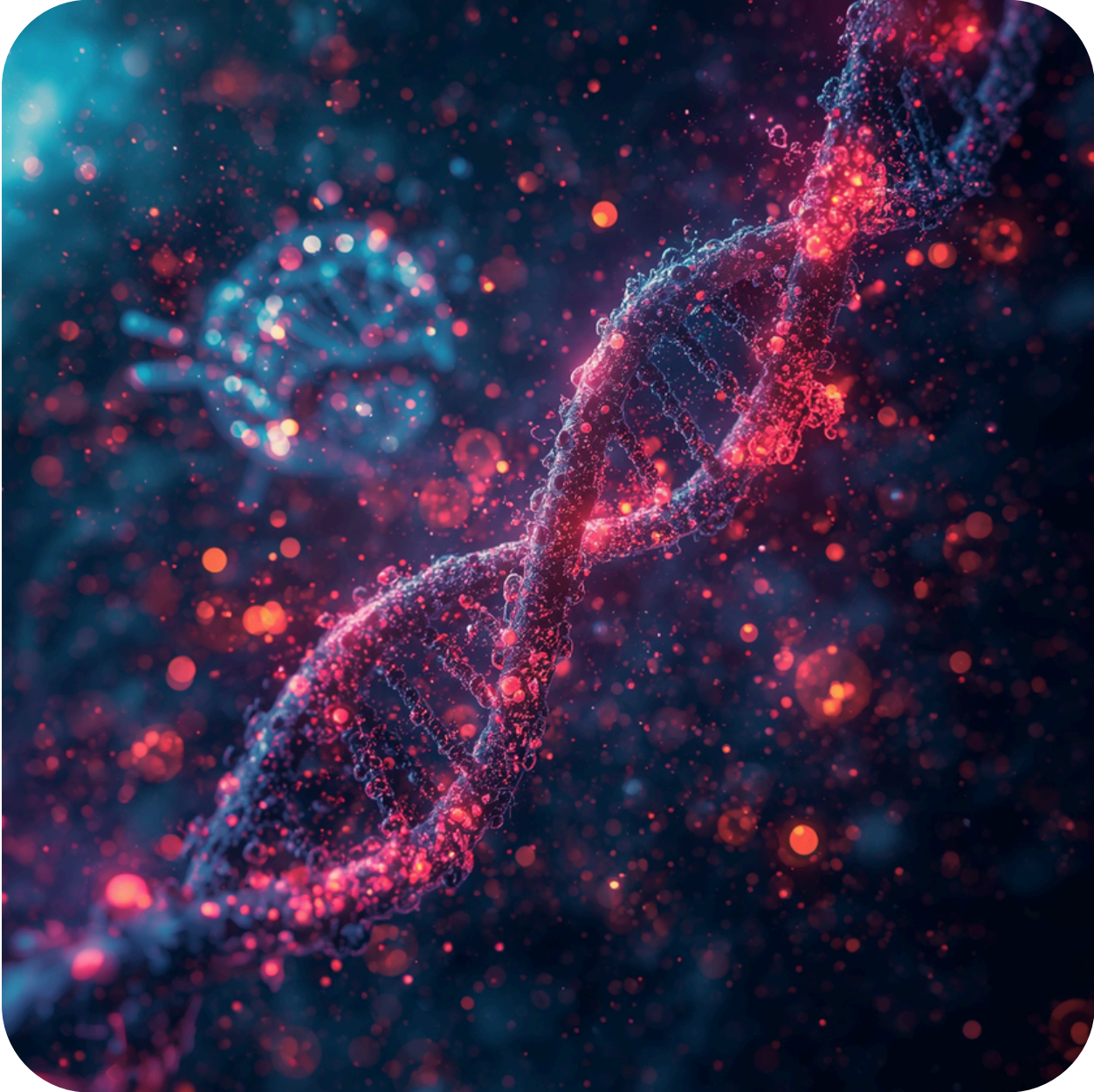


2026 RESEARCH UPDATES



APRIL 30 - MAY 3, 2026
HYATT REGENCY
RESTON, VIRGINIA



NTSAD CONFERENCE | RESEARCH UPDATES & RESOURCES

We have new things planned for NTSAD's Research Day to provide you with opportunities to engage with speakers, researchers, industry partners, and clinicians in a number of ways.

What's New?

- This Research Updates booklet contains the summaries from companies, institutions, and researchers that serve as updates on their work. Some of them are here at the conference.
- For the first time, we have a poster session that gives you the chance to ask questions and better understand the research directly from the scientists. You can refer to the list of the posters that will be presented on Friday, May 1st in between sessions and meals.
- The disease-specific breakouts are structured to allow more time for families to ask questions following the presentations.
- Mark Your Calendars: Following the conference, on Friday, May 15, the Research Team, Valerie Greger, PhD, and Cyndy Perreault-Micale, PhD, will host a Zoom to give you the chance to ask any follow-up questions regarding research.

Finally, these summaries will be shared on the NTSAD Annual Family Conference page on our website following the conference.

POSTER SESSION

Building a Human Variant Database for GM2 Gangliosidoses to Support Families and Providers

Authors: Cintia Carla da Hora, Samuel Kartsonis, Amanda Nagy, Florian Eichler

Clinical Comparison of Gene Therapy-Treated and Untreated Infantile GM1 Gangliosidosis in Siblings

Authors: Hera Akmal, B.S.E.1,2, Precilla D'Souza, DNP1,2, Jean M. Johnston M.S.1,2, Connor J. Lewis, B.S.1,2, Maria T. Acosta, M.D.1,2, Cynthia J. Tifft, M.D., Ph.D.1,2

1 Office of the Clinical Director, National Human Genome Research Institute, Bethesda, MD, USA

2 Medical Genetics Branch, National Human Genome Research Institute, Bethesda, MD, USA

The Infantile Neurodevelopmental Rating Scale (iNDRS): Preparation for Clinical Trials in Tay-Sachs and Sandhoff

Authors: Eileen Gillan, Michael Kiefer, Amanda Nagy, Florian Eichler, Elise Townsend

Nanoparticle Distributed Intravenous Enzyme Replacement Therapy (NanoDIVERT) for Tay-Sachs and Sandhoff Diseases

Author: Jessica Larsen, Noah Arnold, Amanda Gross

Nursing Perspective in the GM2 Clinical Trial – UMass Chan Medical School

Author: Danielle Kokoski, BSN, RN

Affiliation: UMass Chan Medical School, Worcester, MA, USA

SOTERIA Open Label Basket Trial in CLN2, CLN3, Krabbe and Sandhoff Disease

Authors: Lisa Bollinger, M.D., Shrijay Vijayan, PhD., Minsu Kang, PhD

White Matter Injury in Canavan Disease Follows the Spatiotemporal Pattern of Myelination

Authors: Amanda Nagy, Malte Hoffmann, Aliza Wilson, Samuel Kartsonis, Haley McLaughlin, Lizbeth De La Rosa Abreu, Adrian Dalca, Lilla Zollei, Janna Bredow, Annette Bley, Otto Rapalino, Florian Eichler

RESEARCH SUMMARIES

Canavan

CANaspire | Aspa Therapeutics, a BridgeBio Company

This trial is designed to assess the safety and potential benefit of the investigational gene therapy in Canavan patients. Aspa's investigational gene therapy trial uses an AAV9 vector (adeno-associated virus serotype 9), which is designed to deliver functional copies of the ASPA gene throughout the body, including into the brain, to address the underlying cause of Canavan disease. To be considered for participation in the clinical trial a child must have a diagnosis and signs of Canavan disease, be 30 months of age or younger on the expected dosing date, and meet additional study criteria as evaluated by the study doctors.

Developing Human Induced Pluripotent stem cell (iPSC)-based Cell Therapy for Canavan Disease Yanhong Shi, PhD | Beckman Research Institute of City of Hope

My research goal is to find a cure for myelin disorders, especially Canavan disease. My laboratory has been developing human stem cell-based cell therapy for Canavan disease using the induced pluripotent stem cell (iPSC) platform. In this platform, easily accessible cells, such blood cells, can be converted into brain cells that are deficient in Canavan disease patients, therefore, can be used as cell replacement therapy for Canavan disease. We are now performing IND-enabling study and hope to start a clinical trial to test our iPSC-based cell therapy for Canavan disease in a couple of years.

GM2 Gangliosidosis (Tay-Sachs and Sandhoff)

AQNEURSA | IntraBio

AQNEURSA (also known as N-Acetyl-L-Leucine; IB1001; levacetylleucine) has been studied in a Phase IIb clinical trial in adults and children living with GM2 gangliosidosis. In this study, participants received treatment for six weeks followed by a six-week washout period, allowing researchers to compare how patients felt and functioned on and off treatment. The trial met its primary and secondary endpoints with statistically significant results, showing improvements in function such as balance, coordination, fine motor skills, and speech during treatment, with these benefits worsening after the medicine was stopped, suggesting a meaningful treatment effect. AQNEURSA was generally well tolerated, with no serious safety events reported. IntraBio is preparing a resubmission of its application to the U.S. Food and Drug Administration (FDA) for the treatment of GM2 gangliosidosis. We continue to work closely with the National Tay-Sachs & Allied Diseases Association (NTSAD) and global patient organizations and remain committed to advancing our regulatory plans in GM2.

Nanoparticle Delivered Intravenous Enzyme Replacement Therapy (NanoDIVERT) for GM2 Gangliosidosis | Amanda Gross, PhD | Auburn University

The current project for the Gross and Larsen labs is looking at a nanoparticle-based enzyme replacement therapy (ERT) for GM2 gangliosidosis. While this specific project is using animal models for a form of infantile Sandhoff disease, we are also working on treating Tay-Sachs disease with the same polymersomes. Additionally, we have been and are evaluating polymersome based ERT for GM1 gangliosidosis.

Nanoparticle cont'd

The project has been successful in loading of both the Hexosaminidase (Hex) A and B enzymes and β -galactosidase (β gal) enzymes. All three have been active in cell culture. The β gal-loaded polymersomes have been tested in both mice and cat models. The Hex A and B polymersomes have been tested in cells and are about to move forward into mice then cats. While the GM1 and GM2 projects are still in development and optimization in animal models, it is hard to say when they will move to the clinical stage. However, the polymers, or building blocks of the nanoparticles, have been used in other FDA-approved therapies and we are using enzymes that have been studied previously, so we feel the path to the clinical stage might be easier and quicker. Simultaneously, this approach would be an alternative to gene therapy and could be used over a lifespan.

NHRF Gene Therapy for GM2 Gangliosidosis | New Hope Research Foundation

The New Hope Research Foundation (NHRF) is a non-profit operating organization, founded in 2006, with a mission of finding genetic cures for GM2 gangliosidosis and other lysosomal storage diseases that affect the central nervous system. The NHRF gene therapy (NHR01), developed for GM2 gangliosidosis, is intended to adequately restore the deficient HexA enzyme activity that is the root cause for both Tay-Sachs and Sandhoff diseases. The NHR01 gene therapy is designed to deliver a gene vector, a unique sequence of DNA, that enables the body's own cells to express the needed enzyme. Each DNA sequence is packaged in a protein shell, like that of an adeno-associated virus. This shell, similar to an actual virus, allows the packaged DNA sequence to be taken-up by the body's cells.

The NHR01 development program has uniquely addressed five major challenges that have faced gene therapies for GM2 gangliosidosis by:

- Creating a modified HexA enzyme (named HexM) that is more stable and potent and requires only half the total DNA length;
- Packaging the HexM DNA construct within the small AAV protein shell such that it allows more efficient enzyme expression;
- Developing a novel minimally invasive delivery method designed to enhance transport of the gene vector into the brain tissue;
- Addressing the serious immune response that might occur short-term towards the AAV protein shell, and long-term, towards the ongoing expressed enzyme; and
- Efficiently producing a high-quality NHR01 gene vector in sufficient quantities to accommodate the large, intravenous dose required for a human adult.

The NHR01 therapy has been developed in collaboration with world leading scientific and biotechnology experts, and has been shown to be safe and effective in multiple pre-clinical evaluations. A dose response study was conducted with mice bred with a severe form of GM2 gangliosidosis.

Normal mice typically live a little over two years, but mice with this disease survive only about four months. The results of this study were exceptional with mice in the highest dosed cohorts having behavioral assessments and a lifespan indistinguishable from normal mice. Links to our seminal research on the novel enzyme (HexM) gene therapy (Tropak et al., 2015) and other technical papers describing our research can be found at: NewHopeResearch.org/Research.

The batch of NHR01 vector to be used in the first clinical trial is currently in production. Our plan is to initiate a trial in the United States in early 2027. This initial trial will have objectives of first determining the therapy safety, and secondly, stopping or slowing the progression of the disease. The initial study is being planned for adult individuals who are 18 to 65 years of age with verified genetic mutations that are known to result in Late-Onset Tay-Sachs or Sandhoff diseases and who are on a stable medication regimen.

Participation in the clinical trial will involve multiple assessments over time of disease progression including muscle control, coordination, cognition, and verbal communication. Immune suppression will be required for the first three months following gene therapy delivery. Please contact Jack.Keimel@NewHopeResearch.org if you have questions or interest in participating in the clinical trial.

GM2 Gene Therapy 2.0 Update | Heather Gray-Edwards, DVM | UMASS

The gene therapy drug product for Tay-Sachs and Sandhoff disease (AAV9-Bic_HEXA/HEXB) was vialled and is being stored at Mass Biologics GMP facility. Final testing of the drug product is ongoing. The in-life portion of the toxicology study is completed, and we are waiting for the post-mortem pathology report from the 90-day animals. We should have this mid-April. We are working through the Investigational New Drug (IND) modules and plan to have it submitted by the conference. Next steps include 30 days of information requests from the FDA where we answer questions they may have. If they deem it acceptable within this 30-day period, they will approve it. If they require additional information, they will put us on clinical hold until we address all their concerns.

Once the IND is approved, we can move towards institutional review board (IRB) approval. We will use a company for this because they are expedient, and UMass has an institutional conflict because Miguel Sena Esteves is the inventor. Once we have IRB approval, we can start enrolling. Regarding funding, we are teed up to get NeuroNext funding to fund the clinical trial. The next steps with the NIH occur after FDA approval. Enrollment for the trial will be a collaborative effort between Drs. Florian Eichler, Cynthia Tifft, and Eleonora D'Ambrosio. They will attend the conference and can answer questions there.

Development of a Disease-specific Clinical Rating Scale for the Late Onset GM2 Gangliosidoses (Tay-Sachs Disease and Sandhoff Disease) | Julie Kissell | University of Wisconsin

The goal of the project is to develop a GM2 Clinical Rating Scale (GM2-CRS) that is specifically designed for adults with late-onset GM2 and reflects real, lived experiences. The main objective of this work is to create a clear, reliable, and meaningful way to track symptoms and changes over time. This scale is designed to capture information on various aspects of the diseases through neurological exams, patient and observer reported questionnaires, and performance-based tests. The GM2-CRS was developed and improved between 2023 and 2024. Input and feedback from participants and families as well as GM2 experts was used to improve the scale. Now, the scale is being used and tested in an ongoing multicenter natural history study that will continue through 2027. Participants take part in annual study visits and complete homebased assessments and questionnaires at least once per year.

Those who cannot travel can still participate by completing the questionnaires online. This approach helps reduce travel burden while still capturing important information about each condition from a wide range of people. The information being collected helps researchers understand which parts of the scale are most useful and meaningful, and can track change over time. Ultimately, this work is meant to support the GM2 community by improving how disease progression is measured, strengthening future natural history studies, and laying the groundwork for evaluating new treatments in future clinical trials.

Gene Editing for GM2 | Ivakine Lab at The Hospital for Sick Children (aka "SickKids") in Canada

At the Ivakine Lab at SickKids, we are committed to finding a treatment for infantile Tay-Sachs Disease (TSD), focusing on a specific HEXA mutation called c.1278insTATC. Our research involves mice that carry this same mutation and develop symptoms similar to those seen in children with TSD. This enables us to study the disease in depth and test potential treatments effectively. We have already discovered a treatment that greatly extends the lifespan of these mice and alleviates their symptoms, giving us hope that this work could one day help children living with TSD.

A major part of our work is developing gene-editing treatments using CRISPR-Cas9, a technology that allows us to make precise changes to DNA. In simple terms, CRISPR works like a pair of "molecular scissors" that can remove or correct the genetic change causing disease in our mouse model. By targeting the mutation directly, we hope to slow or even reverse the progression of TSD. At the same time, we are also developing an even more precise gene-editing approach called prime editing, which may offer additional benefits in the future.

We are also studying the disease more closely so we can better understand how it affects the body and how to measure whether treatments are working. For example, we have examined the retina in our mouse model to learn more about how TSD causes damage over time. We hope this will eventually allow us to use the retina as a window into the disease and as a way to track treatment effects. Alongside this, we are testing different delivery methods and gene-editing technologies to find the safest and most effective approach. Over the coming years, we hope this research will lead to new therapies that can make a real difference for children with TSD.

SOTERIA Study for Sandhoff Disease | Polaryx

Polaryx Therapeutics is testing PLX200 in children with several lysosomal storage diseases (CLN2, CLN3, Sandhoff, and Krabbe) in the SOTERIA Phase II trial. SOTERIA set up is currently occurring with anticipated enrollment of patients in the second half of 2026.

The study is open label (no placebo, results will be compared against historical control) and looks at safety and preliminary signs of benefit. Progress will be measured using standard rating scales for each disease. Dosing is based on weight to reach drug levels shown to work in preclinical studies, and patients stay on treatment for up to 101 weeks (5 weeks of initial dosing up to the full dose followed by 96 weeks of maintenance).

PLX200 is a repurposed drug already approved by the FDA for treating certain types of high cholesterol and high triglycerides in adults. It is normally taken as a 600 mg tablet twice a day. For this study, it will be given as a flavored liquid twice daily. The drug's safety in adults is well known. Common side effects include stomach upset, abdominal pain, and headaches. More serious risks—though uncommon—include liver problems, muscle issues when combined with some statins, and rare blood-related side effects.

Late onset GM2 Gene Editing | Rick Proia, PhD | NIH

Research in the Proia laboratory is focused on developing a gene-editing approach for late-onset Tay-Sachs disease (LOTS), a condition that usually begins in the teen or adult years and gradually affects movement, strength, and coordination. LOTS is caused by a small change (mutation) in the HEXA gene that prevents brain cells from breaking down certain lipids. Over time, this lipid buildup damages nerve cells.

Our goal is to correct this mutation directly in the brain using a precise form of gene editing called base editing. Instead of adding a new gene, this approach fixes the original DNA mutation. In our recent studies, we delivered this gene-editing system to the brain in a mouse model of LOTS. Even though only a portion of cells were corrected, we saw significant benefits: improved enzyme activity, a large reduction in lipid buildup in the brain, delayed onset of symptoms, and longer survival.

This work is still at an early, preclinical stage, but it provides evidence that correcting the disease-causing mutation could change the course of the disease. With additional research to ensure safety and effectiveness, we hope this approach could move toward clinical studies in the future.

GM2 Gene Therapy | Jagdeep Walia, PhD | Queen's University

The GM2 trial at Queens University hopes to start a clinical trial early next year for infantile and possibly juvenile patients. More information will be shared when it is available.

GM1 and GM2 Gangliosidosis (Tay-Sachs and Sandhoff)

NAVIGATE Study | Azafaros

Azafaros is developing a potential treatment for three rare genetic diseases: Niemann–Pick type C (NPC), GM1 gangliosidosis, and GM2 gangliosidosis, conditions that affect how certain fatty substances are processed in the body, especially in the brain.

Compound

Nizubaglustat is a small molecule that crosses the blood–brain barrier and has a unique dual mode of action. It influences both the production and the elimination of the fatty substance. The drug is administered once daily as an orally dispersible tablet, which dissolves in the mouth or can be mixed with water and given via a G-tube.

Studies

Natural History Study

To understand the natural course of GM1 and GM2, Azafaros conducted a natural history study, PRONTO, to learn more on what happens when there is no interference of any treatment. The published data show the steady decline in patients across age ranges and specifically the impact of the disease on movement (ataxia).

Phase 2 RAINBOW Phase

Thereby, we continued our clinical development path with the Phase 2 RAINBOW study, a 12-week placebo-controlled trial conducted in Brazil in adolescents and adults living with GM2 gangliosidosis or NPC.

Phase 3 NAVIGATE

What we learned from RAINBOW and its extension, directly shaped our current pivotal study, NAVIGATE. The knowledge gained from the RAINBOW program, especially about dosing, safety over time, and biological activity, ensured that NAVIGATE was designed around the measures that matter most to families, clinicians and regulatory agencies.

NAVIGATE includes two sub studies: one for NPC and one for GM1/GM2 gangliosidoses. NAVIGATE is an 18-month Phase 3, placebo controlled, randomized study evaluating whether nizubaglustat can slow or stabilize the progression of these diseases and support day to day functioning. NAVIGATE is already active and enrolling internationally. In the US, several centers are in the process of setting up: California, Dr Hastings, Virginia, Dr Goker Alpan, Minnesota, Drs Dhamija and Jarnes, and Texas, Dr Ramirez.

The goal of NAVIGATE is to determine whether this treatment can meaningfully improve the daily lives of children and young adults living with these rare conditions. As NAVIGATE progresses, it will provide essential evidence to guide future steps toward making a potential therapy available to families affected by NPC, GM1, and GM2 gangliosidoses. For this purpose, ataxia and swallowing are being assessed as well as other manifestations of the diseases that affect daily life like seizure frequency and quality of life.

For more information, please reach out to your health care team or check www.azafaros.com and www.navigate.azafaros.com

Enzyme Replacement for GM2/GM1 Gangliosidosis | JCR Pharmaceuticals

Enzyme replacement therapy (ERT) is a treatment mainstay in many lysosomal storage diseases (LSDs): A functional enzyme is administered intravenously to the individual affected with the disease, finding its way to the lysosome and clearing the accumulated substrate. While about 15 ERTs have been approved for the treatment of lysosomal storage diseases, this approach has shown limited efficacy for the treatment of LSDs with CNS symptoms. The reason is simple: The enzymes are too large to cross the blood-brain barrier and enter the brain.

Enzyme Replacement cont'd

JCR has developed a technology to shuttle enzymes across the blood-brain barrier that led to the commercialization of the first brain-penetrating ERT in Japan (for the treatment of MPS II). Based on this technology, which fuses the enzyme to an antibody that binds a receptor on the surface of the blood-brain barrier, JCR has developed such a fusion protein also for the treatment of GM2 gangliosidosis. The molecule has shown promising activity in animal models for GM2 gangliosidosis and a favorable tolerability and safety profile in early preclinical studies.

JCR, with the support of our partner MEDIPAL Holdings is now developing this molecule towards application in clinical trials. It includes a manufacturing process for the production of clinical material and the conduct of preclinical safety studies that are required before the molecule can enter clinical trials. Eligibility criteria for the clinical trials and a prospective date of trial initiation have not been determined yet.

A similar approach with our blood-brain barrier technology is applicable to other treatments including GM1 gangliosidosis. At this stage, our focus is to advance the fusion molecule for GM2 gangliosidosis into clinical trials as early as possible.

GM1 Gangliosidosis

GM1 Prenatal Gene Therapy | Dr. Tippi MacKenzie | UCSF

Dr. Tippi MacKenzie and her team at the University of California, San Francisco (UCSF) are preparing to open a first-in-human phase I clinical trial of prenatal gene therapy to treat fetuses affected with Type I or Type II GM1 gangliosidosis. The goal of the study would be to understand the safety and efficacy of this treatment when given before birth. Dr. MacKenzie is studying prenatal administration of gene therapy because it has been shown that symptoms of this condition begin during pregnancy, which may limit the effectiveness of therapies given after birth. The experimental therapy that is used has been tested in a clinical trial in children diagnosed with GM1 and was found to be safe, but had limited effectiveness in these patients, likely because the timing of the treatment was late compared to when the disease complications begin.

While this trial would be the first time a gene therapy medicine is given to human fetuses, there has been a lot of work in animal models of fetal therapies supporting the safety of this approach. If Dr. MacKenzie's proposed study is approved by the FDA and the UCSF institutional review board, the drug would be given to affected fetuses through the umbilical vein, between 28-36 weeks' gestation with ongoing follow-up for 5 years after birth.

It is important to the UCSF study team to understand what parents and caregivers of individuals affected with GM1 Gangliosidosis think about this potential study. The UCSF team is conducting an online survey to understand the attitudes. If you are the parent or caregiver of an individual affected by GM1 Gangliosidosis and interested in participating, please share your opinions through the link here: (<https://redcap.link/aeujc3zc>)!

AAV9 Gene Therapy in Type II GM1 Gangliosidosis – A Phase 1-2 Trial | Cynthia Tifft, MD, PhD | NIH Published in the *New England Journal of Medicine*, March 19, 2026

This study looked at a new type of gene therapy for a rare and fatal childhood disease called GM1 gangliosidosis, which affects the brain and leads to gradual loss of skills like movement and communication. Currently, there are no approved treatments. Nine children who had completed 3 years of the study were reported. A total of 15 children have been treated. A single IV infusion of a harmless virus (AAV9) carrying a working copy of the missing gene (GLB1) was administered with the goal of helping their bodies produce beta-galactosidase, the enzyme that is lacking in GM1.

AAV9 Gene Therapy in Type II GM1 Gangliosidosis cont'd

Over three years, the treatment appeared mostly safe, although all children had minimal side effects. The most common issues were temporary increases in liver enzymes (a sign of liver stress), and one child had vomiting that required hospitalization. Importantly, the therapy did what it was supposed to biologically: enzyme levels increased and GM1 ganglioside in the brain (as measured in spinal fluid) decreased, suggesting the treatment was reaching the brain and working at a chemical level.

In terms of how the children functioned, the results were mixed but encouraging. Many children's abilities (like movement and communication) stayed stable or declined more slowly than expected, which is meaningful because, untreated, the disease worsens steadily over time. Brain scans also suggested decrease in the rate of brain atrophy and an increase in the number of white matter tracks, the way the brain forms connections. Overall, while this is an early, small study, it suggests that this gene therapy could help slow disease progression. Further research is ongoing.

The Infantile Neurodevelopmental Rating Scale (iNDRS): Application for Tay Sachs, Sandhoff, Canavan, GM1 and Other Rare Neurogenetic Diseases of Early Childhood

Study team: Eileen Gillan DPT, NCS1, Michael Kiefer PhD, DPT^{1,2}, Amanda Nagy, MD³, Florian Eichler, MD³, Elise Townsend PhD, DPT, PCS1

¹MGH Institute of Health Professions; ²Virginia Commonwealth University; ³Department of Neurology, Massachusetts General Hospital

Overview of our study: Developmental testing used in previous clinical trials involving Tay Sachs and other allied diseases required lengthy evaluations at specialty care clinics and often failed to capture important subtle changes in children's function. To address this concern, the NTSAD funded our group to gather insight from caregivers of children with Tay Sachs and Sandhoff and expert clinicians to develop and pilot a new tool, now called the Infantile Neurodevelopmental Rating Scale (iNDRS). The iNDRS is a brief, 17-item measure that allows a clinician and caregiver to collaborate and complete developmental testing via telehealth, lessening the need for family travel to care centers. The goal of our ongoing iNDRS study is to characterize the natural history of Tay-Sachs and Sandhoff using the scale and validate its use for clinical trials in early-onset neurogenetic conditions, including Tay-Sachs, Sandhoff, Canavan, and GM1.

Work to date: Between January 2024 and April 2026, nine infants (6 Tay-Sachs, 3 Sandhoff) completed Zoom visits with a clinician and caregiver every 1-3 months. Our first paper describing how the iNDRS was developed with the help of the Tay Sachs/Sandhoff community was published in the Journal of Child Neurology in 2024. Next stage findings were presented at rehabilitation and pediatric neurology conferences in late 2024 and 2025. We are now preparing to publish interim study findings from the infantile onset GM2 natural history cohort.

Current opportunities for children and caregivers: We are currently recruiting infants and young children (age 1 month – 5 years old) with confirmed Tay-Sachs, Sandhoff, Canavan, GM1, and other early onset neurogenetic conditions to participate in the study. No clinic visits are required; all participation is via Zoom using a smartphone or laptop webcam. We measure visual, auditory, hand/arm, communication, gross motor, and feeding functions using the iNDRS. Caregivers report their child's symptoms and functional status at the start and with each Zoom visit. This work will help us better understand how children's development changes over time and allow us to prepare the iNDRS for wider use in clinics and clinical trials, which may improve the developmental testing experience for children and their families, and may inform how we conduct and interpret findings from clinical trials.

Contact the study team (mghgm2scaleproject@partners.org) and/or scan the QR code for more information or to enroll:



Visit [NTSAD.org](https://www.ntsad.org) for more research-related information including:

- Biomarker Working Group: secure FDA recognition of GM1 and GM2 ganglioside levels in cerebrospinal fluid as clinically relevant biomarkers for GM1 and GM2 gangliosidosis
- Virtual Biorepository: a resource to facilitate the sharing of research samples, including cell lines and patient samples.
- Scientific Symposium at Annual Family Meeting
- Research Initiative Program: Request for Proposals (RFP): a yearly initiative that enables funding of cutting-edge research to find treatments and cures for Tay-Sachs, Canavan, GM1 gangliosidosis, and Sandhoff diseases. Established in 2002, the program drives our mission forward by funding research that accelerates scientific progress. In addition to grant funding, NTSAD supports researchers by providing letters of recommendation, facilitating scientific connections, and fostering collaboration across the field.
- Scientific Advisory Committee (SAC)
- Late Onset Tay-Sachs and Sandhoff (LOTSS) Think Tank: a select group of researchers and clinicians who convene for a multi-day symposium dedicated to accelerating research toward effective treatments for LOTSS.
- Carrier, Prenatal, and Newborn Screening Initiatives: raise awareness and advocate for comprehensive screening for Tay-Sachs, Canavan, GM1 gangliosidosis, and Sandhoff diseases
- Resources to help patients, providers, and the scientific community:
 - Information for Psychiatric Providers on Treatment of Psychiatric Symptoms in Late-Onset GM2 Gangliosidosis
 - Understanding Therapeutic Approaches - a series of documents to help our community understand the biological basis of the current therapeutic approaches being explored for Tay-Sachs, Canavan, GM1, and Sandhoff diseases
 - Variant (Mutation) Database (VDB) - a resource that can be used to learn more about Tay-Sachs, Canavan, GM1 gangliosidosis, and Sandhoff diseases and many of their associated pathogenic variants
 - Drug Repurposing Papers

**Questions? Want to learn more?
Reach out to the NTSAD Research Team**

Valerie Greger, PhD at vgreger@ntsad.org
Cyndy Perreault-Micale, PhD at c.perreault-micale@ntsad.org



National Tay-Sachs &
Allied Diseases Association