GLIA-CTN ADVOCACY NEWSLETTER

Advocacy Committee Co-Chairs:
Erica Barnes, MA-SLP
Melody Kisor, MS, BCPA
https://theglia.org/glia/advocacy-partners

GLOBAL LEUKODYSTROPHY INITIATIVE | A Rare Diseases Clinical Research Network Consortium

Winter 2025

Have you visited the GLIA- CTN Data Dashboard?

The GLIA-CTN Data Dashboard provides an overview of our multi-center leukodystrophy biorepository, including aggregate data on enrolled participants, diagnoses, sample types, and site contributions. This tool offers a transparent snapshot of the network's reach and activity. It was made possible thanks to the advocacy community's call for accessible data and GLIA's commitment to delivering on that vision.

APPLY NOW:
GLIA-CTN AdvocacyAdvancement Award

APPLY NOW

APPLY NOW

This new award supports leukodystrophy-focused nonprofits in completing six-month projects using GLIA-CTN data and tools. Projects may include developing educational materials, defining burden of disease, expanding dashboards, or preparing for FDA engagement. Two awards are granted annually, with rolling applications and highly recommended pre-application consultations.
Learn more or contact Omar Sherbini at sherbinio@chop.edu

CHOP Names Dr. Amy Waldman Inaugural Elise's Corner Endowed Chair in Alexander Disease Research

Dr. Amy Waldman has been named the inaugural holder of the Elise's Corner Endowed Chair in Alexander Disease Research at Children's Hospital of Philadelphia (CHOP), marking an important milestone for the leukodystrophy community. Made possible through the generosity of the Elise's Corner Foundation together with Jim and Marguerite Pearson, this endowed chair establishes dedicated and sustained support for accelerating research and advancing clinical care for Alexander disease (AxD), a rare and progressive disorder affecting the brain's white matter.
Dr. Waldman is a nationally recognized pediatric neurologist whose leadership has helped shape CHOP into a center of excellence for complex neurological conditions. She founded both the CHOP Leukodystrophy Center and the CHOP Multiple Sclerosis Center, creating two of the nation's leading programs for children with white-matter disorders. As Medical Director of the Leukodystrophy Center, she is known for her deep clinical expertise, her commitment to multidisciplinary care, and her compassionate partnership with families navigating AxD.
With the support of the new endowed chair, Dr. Waldman will expand efforts to better understand AxD progression, establish stronger clinical baselines, identify and validate biomarkers, and create more pathways for earlier diagnosis and emerging treatments. Her appointment signals CHOP's long-term commitment to AxD research and offers renewed hope to families that sustained, focused investment can lead to improved outcomes and a brighter future for children living with Alexander disease. For more information: visit chop.edu

Community Events:

February 23, 2026
FDA Rare Disease Day 2026
Virtual Public Meeting
fda.gov

February 24–26, 2026
Rare Disease Week on Capitol Hill (Advocacy Days)
Washington, DC
https://everylifefoundation.org/rare-advocates/rare-disease-week/

February 28, 2026
NIH Rare Disease Day
https://ncats.nih.gov/news-events/events/rdd

March 24th, 2026
Awareness Day for LCC Foundation
https://thelccfoundation.org/

May 23, 2026
The 2026 TUBB4A Family Conference
Philadelphia, PA
https://www.h-abc.org/

July 22-25, 2026
The LCC Foundation Conference
St. Louis, MO
lccfoundation.mo@gmail.com

June 25-27, 2026
United Leukodystrophy Foundation Family Conf
Chicago, IL
https://ulf.org/news/conference/

WELCOME

Welcome Dean Karavite!

Please join us in welcoming Dean Karavite to the GLIA team. Dean has joined us as Principal Human–Computer Interaction Specialist and will lead efforts to understand user needs as we develop an MLD research website and other resources across the GLIA-CTN. As part of his role, Dean has been interviewing patient advocacy groups to learn what information and features matter most to families and communities. We're excited to have his expertise guiding a user-centered approach to our work.

Advocate Spotlight : Ashley Dike

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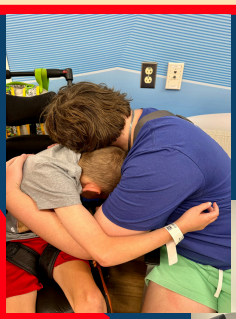


Top Photo:
Zac and Ben
Right Photo:
Dr. Jamie Fraser with Ben
Photo Below:
Ashley (Mom), Danny (Dad), Zac, Eli, Ben, Sam, and Patrick Mahomes at Ben's Make a Wish!

Share your passion for the work you do:
My passion is deeply personal. I'm a parent to two sons diagnosed with LCC. That experience drives my commitment to ensuring families are supported in their present circumstances while also building a lasting legacy of progress. I'm motivated by the belief that no family should feel alone—and that through collaboration across the leukodystrophy community, we can create hope not only for today's patients, but for future generations, including my grandchildren and great-grandchildren.

What areas of advocacy are you most passionate about?
Patient-centered research infrastructure, especially patient registries, and ensuring that collected data is used effectively—through analysis, publication, and creating meaningful impact for patients and families.

What advice do you give to someone just starting in rare disease advocacy?
Start small, listen often, and build relationships early. Collaboration and sustainability matter more than doing everything at once.

What's one project or moment that you're especially proud of?
Helping lead and host a Patient-Led Listening Session with the FDA in July 2025 was a meaningful milestone. It reflected years of community-building and collaboration, and ensured that patient and caregiver voices directly informed regulatory conversations—while reinforcing the importance of using patient-generated data to drive real progress.

What is something that people might not realize about this work?
Rare disease advocacy requires learning many disciplines at once—business, psychology, science, medicine, and clinical practice—often without formal training. Most advocates are also caregiving for loved ones, carrying both profound pain and deep purpose at the same time. It is demanding, emotional work, but it is also a role filled with meaning—and one I wouldn't choose any other way.

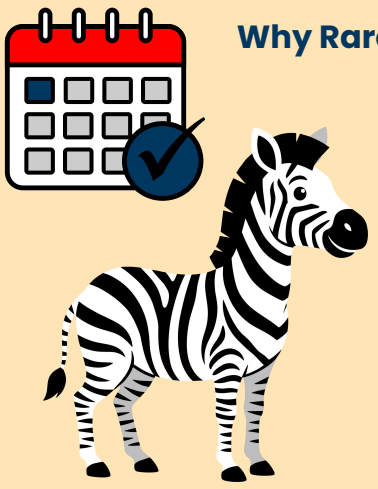
NEW

CureARS Launches New Patient Registry

CureARS, representing all 19 mtARS genes, is excited to announce the launch of the MAPS Patient Registry in partnership with Nord. This new registry is designed to empower patients and families, advance research, and strengthen the collective understanding of mtARS-related conditions.
By participating, individuals can help accelerate research efforts, improve awareness, and support the development of future treatments.
Learn more and join the registry here.

February 28, 2026

Why Rare Disease Day Matters for the Leukodystrophy Community



Rare Disease Day, observed each year on the last day of February, is a global moment to raise awareness, build understanding, and strengthen advocacy for rare conditions. For the leukodystrophy community, it is an important opportunity to come together and ensure our voices are heard.
This day underscores the ongoing need for earlier diagnosis, expanded screening, better access to specialized care, and sustained investment in research and clinical trials. It also offers time to recognize the resilience of individuals living with leukodystrophy, the dedication of caregivers, and the commitment of clinicians and researchers working toward meaningful progress.
There is no single way to participate. Families may choose to share their story, wear purple, attend an event, educate others, or pause to reflect. **We invite you to share how you are recognizing Rare Disease Day**—through advocacy, awareness, or a personal moment. Each shared experience helps amplify our community and move leukodystrophy care and research forward.
Together, rare is powerful.
To learn more visit: https://www.rarediseaseday.org/

Celebrating 10 years of progress toward the treatment of GM1 gangliosidosis: *An update from the Cure GM1 Foundation*

The Cure GM1 Foundation was founded in 2015 to raise awareness of GM1 gangliosidosis and advocate for potential treatment pathways for the disease. In the past 10 years, Cure GM1 has made tremendous progress in laying the groundwork for new treatments to emerge. Below is a brief overview of Cure GM1's work and its multi-pronged approach toward finding hope for GM1 families.

What is GM1 gangliosidosis?

GM1 gangliosidosis is an ultra-rare, inherited genetic disorder that primarily affects children. It causes debilitating and progressive declines in the areas of physical and mental development, mobility, vision and other neurological functions. There is currently no cure or effective treatment.

GM1 is an LSD, similar to Tay-Sachs and Niemann-Pick. An LSD is a condition in which a "defective gene" causes the absence or deficiency of cellular enzymes needed to clear toxins from the body. This in turn leads to toxic buildup, eventually causing irreversible damage to the brain, spleen, liver and other organs.

How is the Cure GM1 Foundation working to improve outcomes for GM1?

Diseases like GM1 are complex, and the Cure GM1 team, led by GM1 parent and patient advocate Christine Waggoner, recognizes that its eventual treatment will be just as complicated. The Cure GM1 Foundation has spread a wide net in fighting the disease.

The organization supports:

Gene therapy/small molecule treatment: In its first decade, Cure GM1's work has resulted in multiple adeno-associated virus (AAV) gene therapy trials for GM1, as well as several small molecule clinical trials. Within its first four years, Cure GM1 had already helped bring the first clinical trial for GM1 to fruition: NIH Intravenous AAV9. The trial is open to enrollment and has treated 17+ trial participants to date. In addition, Children's Hospital of Philadelphia is a site for the Passage Bio gene therapy trial. The study focused on finding a safe dose of AAV gene therapy in children with early infantile or late infantile GM1.

Most recently, the first patient was dosed in a global Phase 3 clinical trial for a small molecule. Led by Azafaros, the trial explores the use of Nizubaglustat as a potential treatment option for GM1 and other rare lysosomal storage disorders. FDA approval is possible pending further study.

Enzyme Replacement Therapy (ERT): ERT is a tried-and-true approach that addresses the root cause of GM1. Decades of research with drugs like Aldurazyme and Brineura could offer a roadmap for using ERT for GM1. Thus far, Cure GM1 has invested more than $750,000 in ERT efforts and plans to expand that investment to millions.

Early diagnostics: The treatment window for several promising AAV-based gene therapy trials is narrow. Many are diagnosed too late to benefit. To help, Cure GM1 has also funded the optimization of the first-ever newborn screening assay for GM1.

Cure GM1 Foundation founder Christine Waggoner, with daughter Iris, age 17

Iris, age 5 at diagnosis

2025 Annual GM1 Community Conference

Data acquisition: Cure GM1 supports natural history data sharing and the largest patient registry for GM1 globally. The Cure GM1 census and registry aims to examine patient population data key to drug development.

Community: The organization's Annual GM1 Community Conference brings together families, researchers, and biotech companies to champion positive outcomes for those living with GM1. Cure GM1 also leads monthly family support chats.

Partnership: Lastly, the Cure GM1 Foundation recognizes that progress requires partnership. Cure GM1 organized the first-ever patient-focused drug development meeting with the FDA and has also championed accelerated approval for ultra-rare disease treatment based on biomarkers.

Treatment for those living with GM1 gangliosidosis has historically been nearly non-existent. With the Cure GM1 Foundation's multi-level, multi-pronged approach, meaningful progress is being made.

To learn more, visit **curegm1.org.**

Contributed by Jess Madison, CUREGM1

NORD Webinar Recording Now Available: Open Enrollment for Rare Disease Families

The National Organization for Rare Disorders (NORD) recently hosted a webinar titled "Stay Covered: Open Enrollment Essentials for Rare Disease Families," and the full recording is now available to watch online. The session covered key topics such as understanding open enrollment, comparing health insurance plans, navigating recent insurance changes, and tips for the appeals process. NORD's Policy Team, along with Jennifer Shumsky of JLS Consulting, shared practical guidance designed to help rare disease patients and families maintain access to needed care and medications. Additional resources, including a webinar FAQ and NORD's Claim Your Care program, are available to support families in navigating health coverage decisions.

Watch here: youtube.com

GLIA-CTN Advocacy Quarterly Meeting Recap (December 15, 2025)

- Advocacy leaders highlighted strong collaboration across work groups and meaningful engagement with partners throughout 2025.
- In 2026, advocacy efforts will expand with continued partner collaboration and new opportunities for member involvement.
- Active advocacy work groups include Patient Registry, FDA Engagement, and Special Projects, and members are encouraged to participate in one or more groups.
- A new advocacy work group is being explored to support publications, conference abstracts, and presentations.
- Policy updates were shared from the Rare Disease Congressional Caucus, including early planning for PDUFA 8 and increased attention to Scientific Focused Drug Development (SFDD) meetings alongside Patient-Focused Drug Development efforts.
- Participation in upcoming leukodystrophy-specific and broader rare disease conferences was discussed, with details to be shared as plans are finalized.
- The meeting concluded with appreciation for advocacy leaders and volunteers, with special recognition given to Melody Kisor and Erica Barnes for organizing and leading our advocacy efforts throughout the year.

Progress in Rare Disease: Major Wins for the Leukodystrophy Community

written by: Lesa Brackbill

While 2025 presented new challenges in the Rare Disease and Newborn Screening (NBS) spaces, the leukodystrophy community has achieved significant victories.

- Krabbe Disease: We celebrate the addition of six new states implementing Krabbe screening this year, bringing the national total to eighteen. Looking ahead to 2026, we anticipate that at least eleven more states will adopt Krabbe disease screening.
- Metachromatic Leukodystrophy (MLD): MLD has been officially added to the federal Recommended Uniform Screening Panel (RUSP), marking a major milestone for the rare disease community and a significant step forward for early detection and treatment **(More information here)**
- Cerebrotendinous Xanthomatosis (CTX): The recent approval of a new treatment for CTX is a strong catalyst. We expect this development to drive rapid expansion of NBS for CTX in the near future.

Advancing Newborn Screening takes teamwork. If you are an advocate looking to expand NBS for your specific condition, I would be delighted to share the insights and guidance I've gained through my work. Please reach out—let's discuss how we can partner for success!

Want to join a PAG Workgroup?

Email stoneys@chop.edu to join!

Patient Registries
Led by: Ashley Dike
Meets **first** Wednesday of every month
11 AM EST Virtually

Engaging with the FDA
Led by: Erica Barnes
Meets **second** Wednesday of every month
11 AM EST Virtually

Advocacy Driven Special Projects
Led by: Melody Kisor & Storm Greenway
*Meets **third** Wednesday of every month*
11 AM EST Virtually

Erica Barnes Recognized by Senator Amy Klobuchar

At the National Organization for Rare Disorders (NORD) 2025 Breakthrough Summit, **U.S. Senator Amy Klobuchar**, co-chair of the bipartisan Rare Disease Congressional Caucus, recognized Erica Barnes for her leadership and dedication to the rare disease and leukodystrophy community. Speaking to advocates, researchers, and families, Senator Klobuchar reaffirmed her commitment to rare disease policy, emphasizing the importance of lived experience in driving progress, a central theme of the summit titled "From Voices to Breakthroughs."

Alexander Disease Community Updates

End Alexander Disease PFDD Meeting

On December 5, 2025, End Alexander Disease hosted a virtual Externally-Led Patient-Focused Drug Development (EL-PFDD) meeting, giving community members a rare opportunity to share their lived experiences with Alexander disease directly with regulatory stakeholders, researchers, and advocates. Participants discussed daily impacts of the disease, current treatment challenges, and priorities for future therapies. Feedback from this meeting, including survey responses and public comments, will be included in a Voice of the Patient Report to help inform research and regulatory decision-making. Community members can now watch the replay of the AxD EL-PFDD meeting, and public comments will continue to be accepted through **January 5, 2026**. Watch the live stream here: https://www.endaxd.org/endaxdel-pfdd

Zilganersen Progress

In a major clinical milestone, zilganersen (ION373) demonstrated statistically significant and clinically meaningful improvements in gait speed, as measured by the 10-Meter Walk Test, in a pivotal Phase 1–3 trial in people living with Alexander disease. This marks the first time an investigational therapy has shown potential disease-modifying impact for this rare condition. Favorable safety results were also reported, and the developer plans to submit a New Drug Application (NDA) to the U.S. Food and Drug Administration in the first quarter of 2026. (Source: ionis.com)

Research Updates

Gavazzi, F., Martin, A., Sevagamoorthy, A., Vaia, Y., Vincent, A., Woidill, S., D'Aiello, R., DeMauro, S. B., Lorch, S. A., Vanderver, A., & Adang, L. A. (2025). Experiences and hope in caregivers of children with Aicardi Goutières syndrome. Journal of Child Neurology, 40(7), 543–554. Article Link

This qualitative study interviewed caregivers of children with Aicardi–Goutières syndrome to map emotional, practical, and informational challenges across the disease course. Caregivers described intense grief, chronic uncertainty, and frequent care coordination burdens, but also sources of resilience including peer support, advocacy, and hope driven by emerging research. The authors highlight key unmet needs—mental health supports, clearer prognostic information, streamlined multidisciplinary clinics, and caregiver-inclusive research priorities—and recommend family-centered models that integrate psychosocial services with clinical care.

Page, N., Nagy, A. M., Eichler, F. S., & Ream, M. A. (2025). Seizures in childhood cerebral adrenoleukodystrophy. Developmental Medicine & Child Neurology, 67(8), e134. Article Link

This clinical report details seizure phenotypes, timing, and management considerations in children with cerebral X-linked adrenoleukodystrophy (cALD). The series characterizes common seizure types (focal and generalized), their relationship to MRI-demonstrated disease progression, and typical responses to antiseizure medications. Authors underscore the need for heightened surveillance for new-onset seizures as a manifestation of cerebral involvement and recommend individualized anticonvulsant strategies and integration of seizure management into broader neuro-oncologic and transplant care pathways.

Srivastava, I. N., et al. (2025). Differentiating incidental from pathologic brain MRI findings in asymptomatic boys with X-linked adrenoleukodystrophy: A multicenter study. Neurology, 105(10), e214321. Article Link

In this multicenter imaging study, researchers analyzed brain MRIs from asymptomatic boys with X-ALD to distinguish incidental white-matter variations from early, pathologic demyelinating changes. They report specific radiologic features and risk predictors (distribution, lesion evolution, contrast enhancement patterns) that increase the likelihood an abnormality reflects presymptomatic cerebral disease. The findings support refined surveillance imaging criteria to improve timing of referral for interventions such as hematopoietic stem cell transplant while reducing unnecessary alarm and interventions for benign incidental findings.

Gavazzi, F., et al. (2025). Critical functional domains in pediatric-onset TUBB4A-related leukodystrophy: A clinical and caregiver's perspective. Pediatric Neurology, 173, 156–165. Article Link

Through clinician assessments and caregiver-reported outcomes, this mixed-methods study identifies the functional domains most affected in pediatric-onset TUBB4A-related leukodystrophy and those prioritized by families. Motor function (gait, tone, coordination), communication, feeding and swallowing, and mobility/positioning emerged as consistently high-impact areas. The authors propose these domains as candidate endpoints for clinical trials and emphasize aligning care plans and outcome measurement with caregiver-identified priorities to ensure meaningful benefits in daily life.

Gavazzi, F., et al. (2025). Functional ability profiles in beta-propeller protein-associated neurodegeneration (BPAN). Molecular Genetics and Metabolism, 146(3), 109253. Article Link

This descriptive cohort study maps patterns of motor, cognitive, and activities-of-daily-living impairment across ages and disease stages in BPAN. The paper documents heterogeneity—some individuals maintain ambulation and communication into adolescence while others show early, progressive decline—and identifies common trajectories useful for prognostic counseling. The authors recommend standardized functional profiling to guide individualized care, anticipatory planning, and selection of clinically meaningful endpoints for future therapeutic trials.

Dominguez Gonzalez, C. A., et al. (2025). Reporting ABCD1 variants as actionable secondary findings on exome and genome sequencing. Genetics in Medicine, 27(7), 101425. Article Link

This policy-oriented analysis evaluates whether ABCD1 variants (causative of X-linked adrenoleukodystrophy) meet criteria to be reported as actionable secondary findings in genomic testing. The paper synthesizes evidence on clinical actionability (screening, surveillance, preventive interventions), variable age of onset and penetrance, and ethical implications for family members. The authors offer practical recommendations for laboratories and clinicians on variant classification, informed consent, cascade testing, and counseling strategies aimed at maximizing early detection of cerebral disease while minimizing undue harm from uncertain findings.

Gavazzi, F., et al. (2025). Exploration into lived experiences of multiple sulfatase deficiency–affected individuals and their families. Journal of Child Neurology, 40(10), 852–861. Article Link

Using qualitative interviews, the authors explore the experiences of individuals with multiple sulfatase deficiency and their families, focusing on diagnostic journeys, everyday care demands, and psychosocial impact. Recurrent themes include prolonged diagnostic odysseys, complex multisystem care needs (neurologic, orthopedic, feeding), caregiver burnout, and the centrality of community and advocacy groups. Participants called for improved diagnostic pathways, coordinated care teams, access to rehabilitation and palliative supports, and meaningful engagement of families in research priority setting.

Weiß, M., et al. (2025). Deep intronic SVA_E retrotransposition as a novel factor in Canavan disease pathogenesis. Human Gene Therapy, 36(17–18), 1248–1256. Article Link

This molecular genetics study identifies a deep intronic insertion of an SVA_E retrotransposon in the ASPA gene as a previously unrecognized cause of Canavan disease in affected individuals. The insertion disrupts normal splicing and ASPA expression, expanding the mutational spectrum beyond exonic variants and large deletions. The authors emphasize the diagnostic implications—advocating for inclusion of intronic and structural variant analysis when standard testing is negative—and note potential avenues for targeted molecular therapies or antisense strategies that could restore proper splicing.