

DLG4 SHINE Foundation

2025

ANNUAL IMPACT REPORT



Mission & Projects

Our mission is to improve the quality of life for children and families impacted by DLG4 SHINE through supporting collaborative research among international medical professionals, nurturing a family community, and raising worldwide awareness.

Advocacy Projects

	Details	Outcomes
ICD-10 Code	<ul style="list-style-type: none">Continued to advocate alongside other disease groups	<ul style="list-style-type: none">Finally obtained a unique ICD10 code for DLG4-related synaptopathy in September
Inaugural Conference	<ul style="list-style-type: none">Held in New Jersey on July 17th and 18th	<ul style="list-style-type: none">Gathered over 100 community members and researchers/clinicians

Community Support Projects

	Details	Outcomes
Inaugural Scientific and Family Conference	<ul style="list-style-type: none">Held scientific presentations, sample collection, and community events in Rahway, New Jersey on July 17th and 18th	<ul style="list-style-type: none">Gathered over 100 community members and researchers & clinicians, including 17 affected individuals
Live scientific webinars	<ul style="list-style-type: none">Three live, recorded, expert-led virtual scientific webinars	<ul style="list-style-type: none">Appreciation from the community about the opportunity to hear from experts and the quality of the information
Medical Considerations Document	<ul style="list-style-type: none">Dozens of documents sent to patient families in 3 languages	<ul style="list-style-type: none">Ongoing requests for the document and positive comments about its value with medical care teams.

Mission & Projects

Research Projects

Details

Outcomes

AAV9

- AAV9 projects results presented

- Showed promising proof of concept for AAV9 treatment efficacy for DLG4 SHINE

Small Drug Repurposing Screen

- Small drug repurposing screen results presented

- Evidence of efficacy of high doses of DHA warranting progressing towards a clinical trial

mRNA-based Drug Screen

- Assessment and start of a novel approach

- Collected and sent samples to start analysis

Mechanism of action research

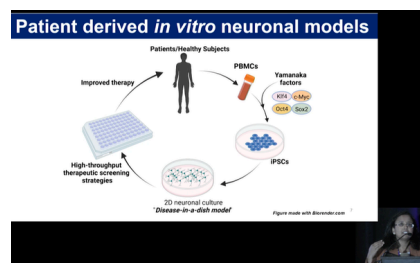
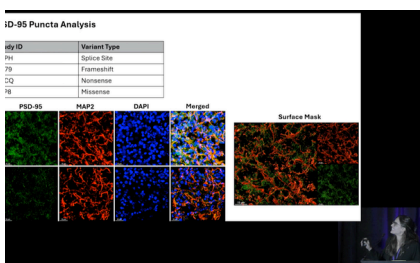
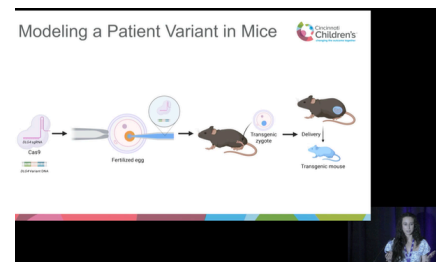
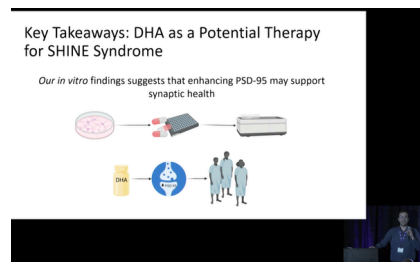
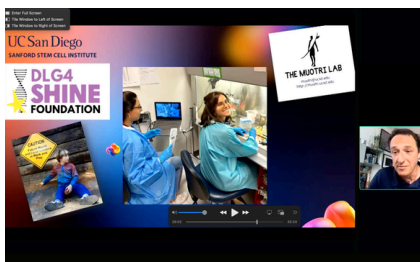
- Start of a project to study the mechanisms of action of splice-site variants

- iPSCs of a splice-site variant and a control have been developed and are being characterized

Biorepository

- Expanded the number and variety of bio-samples and animal models for DLG4

- Sponsored 2 more mice models and collected blood samples and nasal swabs for over 20 patient/control pairs



THE YEAR IN REVIEW



At the very start of 2025, the board finally committed to holding a conference. We had been talking about the possibility for two years. But with exciting science happening in multiple areas of interest to our population and close to yielding results, we knew it was the right time to gather everyone. More than half the year was therefore dedicated to the planning, holding, and wrapping up of this very important event. The conference also allowed us to strengthen the mutually beneficial relationships with both Simons

Searchlight and COMBINEDBrain. And while this was a very time intensive project, we were able to also create relationships with new researchers and start or plan new projects in complementary areas which could offer additional therapeutic pathways, refined diagnostic accuracy, and clinical endpoints for future clinical trials. The news of finally obtaining an ICD10 code, the result of years of advocacy, was the cherry on top of what feels like our most successful year yet.

Seasonal Highlights

WINTER 2025

After a successful 2024 end-of-year fundraising and with projects coming to fruition, the board decided it was finally time to commit to organizing a scientific and family conference and actively started planning the event.

SUMMER 2025

Time and energy were entirely devoted to the inaugural DLG4 SHINE Scientific and Family Conference. Held in Rahway, NJ, it gathered about 20 families and a dozen researchers and clinicians, coming from 7 different countries. It was a resounding success and fostered a spirit of collaboration and community among families and scientists alike.

SPRING 2025

While busy organizing the conference, the board assessed and shaped 4 cutting edge research projects, two of which are now underway, one about to start, and the third in final steps of drafting the agreement.

FALL 2025

With past projects showing promising results for potential therapies and new projects about to start, the board consulted scientific advisors to best select priorities and strategize next steps for the foundation. The end of the year was focused on fundraising for the ambitious plan ahead in 2026 and beyond.

THE HIGHLIGHT OF THE YEAR

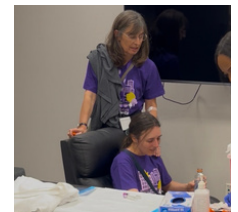
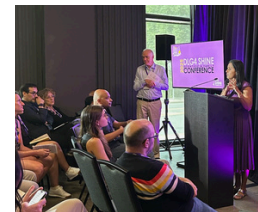
The inaugural

DLG4 SHINE SCIENTIFIC & FAMILY CONFERENCE

Held over two days, this event was the perfect marriage of science and community. A dozen researchers from 4 countries, 2 research consortium partners, over 20 DLG4 SHINE families from 5 countries, and 17 DLG4 SHINEing Stars gathered to learn from and bond with each other. The highly positive feedback from all participants and the smiles on the SHINEing stars faces leads us to believe the event was a success.

The first day was a very full agenda of scientific presentations to allow the researchers to share and the families to learn about all the incredible progress we are making towards therapeutics. And in a joint effort with Simons Searchlight and COMBINEDBrain, we also were able to collect priceless biosamples from affected SHINEing Stars and controls to enrich our biorepository.

Research & Blood Draw



SHINE Camp



While their parents and caregivers were learning about scientific knowledge and advancements, the foundation was thrilled to offer SHINEing Stars the rare opportunity to get together and have fun at a day camp. With the help of the amazing team at Kids Corporate Events, they enjoyed a Beach Party Bonanza day, tailored especially for them, during which they did crafts, played games, and watched a movie.

Sib Shop & Bonding

The conference created yet another unique opportunity: for DLG4 SHINE siblings to meet. They were able to share their experiences and express their feelings with each other privately during a Siblings Workshop. They shared them again, this time with the world during a filmed interview aimed at shedding light on their unique perspective. And most importantly, they were able to simply bond with each other over the two days of the conference.



Community Building

Opportunities to meet are few and far between for members of a rare disease community! These two days offered that chance to bond to the many subgroups that make a community: children (affected and siblings a like), dads, moms, full families, and even the board members, most of whom had never met in person!



Family Day & Dance Party

The last afternoon was a fun family-focused summer fair. Activities included balloon artists, face painters, a wildlife show, Italian ice treats, and a magician. We then ended on a high note with a pizza dinner and the Our Night To SHINE dance party, with a DJ and a photo booth.



THE DLG4 SHINE COMMUNITY

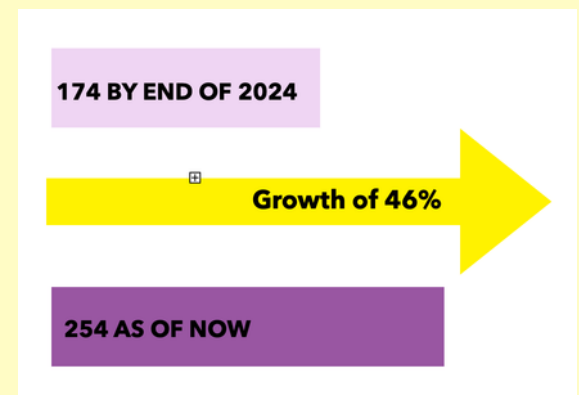
AT A GLANCE

To date, we are aware of about 350 cases of DLG4-related synaptopathy worldwide.

Of those, 254 individuals or their caregivers have registered with our foundation and shared some medical and demographic details about themselves and their loved ones. This allows us, among other things, to know where around the world DLG4 SHINE can be found.

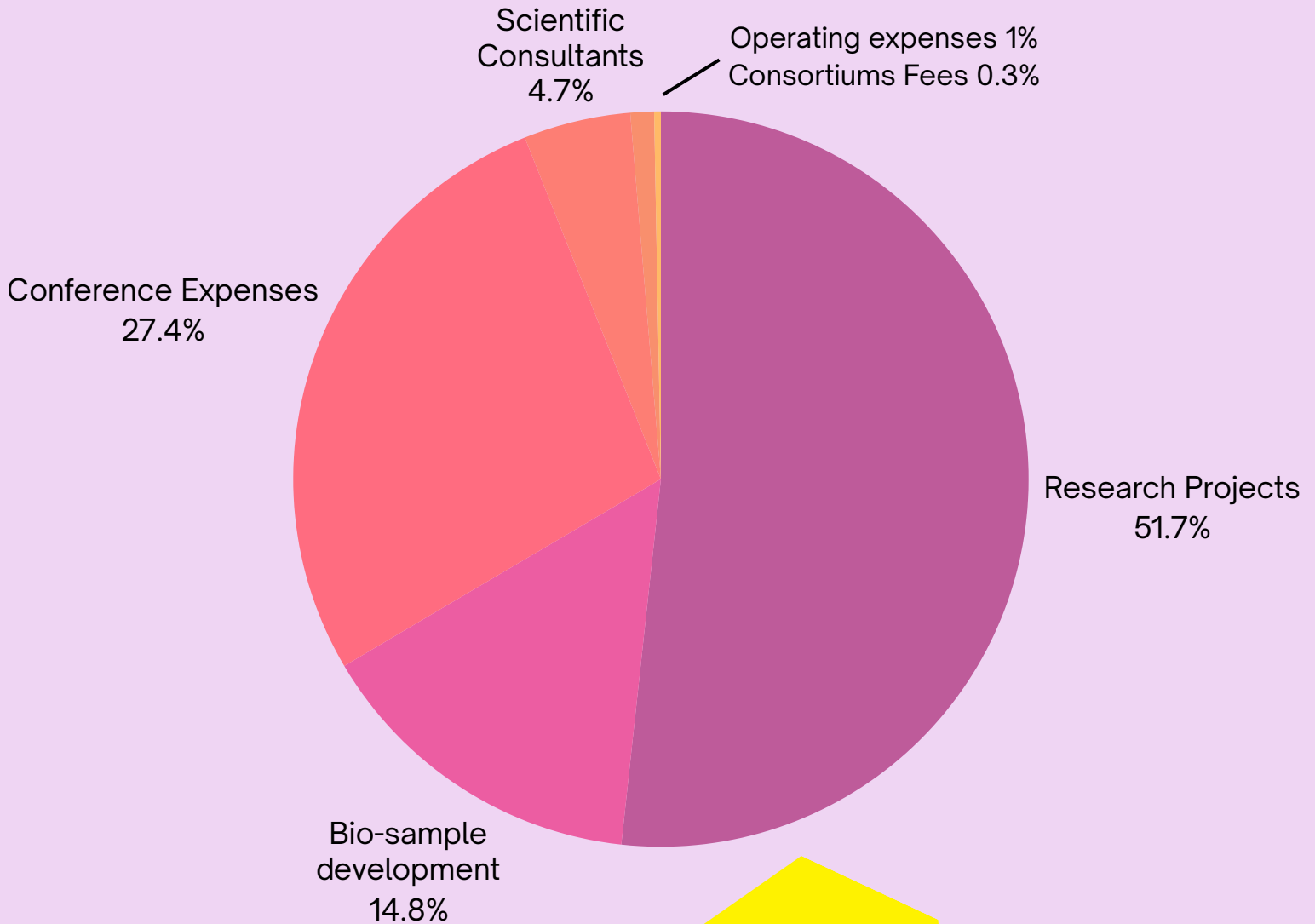


The census at the end of 2024 included 174 families who had registered with the foundation, which means there has been an incredible 46% growth since then.



DLG4 SHINE FOUNDATION FINANCES

The DLG4 SHINE Foundation is dedicated to driving science for new therapeutics, supporting community, and ultimately finding a cure for every person affected by a DLG4 variant. In 2025, our grassroots fundraising efforts yielded **\$446,444** or **126.5% more** than the previous year.



*** These numbers are estimates. Revenues include payments made directly to research centres on behalf of the Foundation. Numbers above will be finalized. The tax return will be available on our website at dlg4shine.org.



**OUR WORK WOULD NOT BE
POSSIBLE WITHOUT THE
GENEROUS SUPPORT OF OUR
DONORS.**

**WE ARE AN ALL-VOLUNTEER BOARD AND
DONATIONS DIRECTLY FUND OUR RESEARCH,
ADVOCACY, AND COMMUNITY PROGRAMS.
THANK YOU FOR SUPPORTING OUR MISSION.**

ACKNOWLEDGEMENTS

Thank you so much to the entire board of the DLG4 SHINE Foundation for their dedication to our mission, to the DLG4 SHINE families who hosted fundraisers or participated in various efforts and initiatives, and to our Medical Advisory Board for their continued guidance and support.



dlg4shine.org