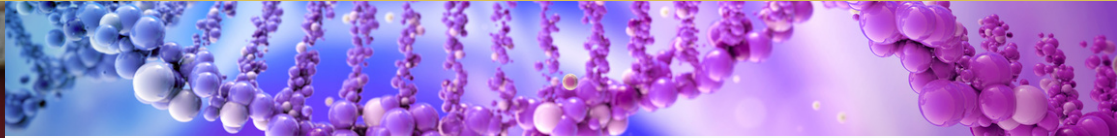


SHINE SYNDROME FOUNDATION NEWSLETTER



SHINE NEWS AND A THANK YOU!

As we approach the one-year anniversary of the SHINE Syndrome Foundation, we want to thank you for your support and for believing in us as we live out our organization's mission statement. You have helped make a difference! Because of our supporters' generosity, we recently paid a full year's salary for 2023 for Amanda Levy to continue her work on DLG4 at the University of Copenhagen. Amanda regularly meets with our board to update us on progress. She is currently working with synthetic DNA vectors to study the functional mechanism of missense variants. We could not have done this without you!

Thank you for your continued support as we help advance research and build a community for all DLG4 families! Interacting with and reposting public social media posts, participating in research opportunities, and sharing fundraisers were all key parts of our successful first year. We recently crossed the \$100,000 threshold for the year and are so grateful. THANK YOU!

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CORDS REGISTRY

This month, we are introducing our second patient registry. The goal of Simons Searchlight is to compare DLG4 variants to *other* neurodevelopmental disorders with the use of validated surveys. The goal of the CoRDS registry is to get a very specific look at DLG4 variants and the things that make a SHINE diagnosis unique. This registry was created entirely by SHINE parents.

To participate in this registry, see enrollment information on our website, <https://shinesyndrome.org/wp/cords/>, and in our private family FB group. As a completion incentive, the first 20 families who complete the survey will receive an international VISA gift card with a \$100 USD value. We will also hold a raffle for an Apple Watch 8 for all families who complete the CoRDS registry by **December 31, 2022**. All incentives are provided by the SHINE Syndrome Foundation but Sanford Health will be responsible for conducting the raffle and distributing the incentives.





SHINE MEET THE MEMBERS

WHO'S WHO?

Marie-France Gervais, or Mara for short, is the SHINE mom of Cédric. She is one of our founding board members and current VP. Mara lives in Ottawa, Canada with her husband and SHINE son, Cédric.



MARIE-FRANCE GERVAIS
Vice President, SHINE Syndrome Foundation

The SHINE Syndrome Foundation recently joined COMBINEDBrain to collaborate on research opportunities within the DLG4 community as well as other rare diseases that fall into similar categories. Mara will serve on the board for the SHINE Syndrome Foundation.

Our board is comprised of seven members who have worked tirelessly over the last year to support our mission: to improve the quality of life for children and families impacted by SHINE Syndrome/DLG4-related synaptopathy through supporting collaborative research among international medical professionals, nurturing a family community, and raising worldwide awareness.



MEET SHINE STAR, BENTLEY

Bentley is a loving, sweet, and funny boy. He is 8 years old and lives with his family in Wyoming state, USA. He has a smile that will light up the room. He enjoys playing with his kitchen set and baby dolls (as long as his sisters play). Mac 'n cheese and sausage are his all-time favorite foods. Bentley was wrongfully diagnosed at 2, with EDS. We started seeing a new PCP in 2021 and she got us on the right track to get a correct diagnosis for him. He is newly diagnosed with SHINE, ESES, and CSWS.

HOW TO SHINE GUIDE

The How-to SHINE guide will be housed on our website indefinitely. Please visit the SHINE Syndrome Foundation website at the address: www.shinesyndrome.org to view the latest edition of the document, as it is updated occasionally. We plan to roll out versions of the guide in other languages in 2023.

2023 SHINE FOUNDATION BOARD

The SHINE Syndrome Foundation will be holding our annual elections in January with new terms beginning in February. If you are interested in joining us, email Laura stating your interest at lpalmer@shinesyndrome.org by 1/6/23. Also plan to attend our January board meeting, which is scheduled for Sunday, January 8th at 12:00 EST. Election information will be sent out prior to the January 8th meeting.

SAMI CAMERA PARTNERSHIP

The SAMI camera is an epilepsy monitoring device for nighttime. It works by using an infrared camera to monitor your child and alarms at settings predetermined by the family. Use the code SHINE10 to get 10% off a SAMI purchase. For more information about the camera, please visit the SAMI website, our website or the parent FB page in the files section.



MICE MODELS

Our first DLG4 mouse model, a frameshift variant, is underway at Cincinnati Children's Hospital Medical Center (CCHMC) in one of the research labs. CCHMC plans to study the sleep and epilepsy characteristics of this disorder. They expect to have enough mice generated to characterize in about six months (late April-May). A couple of other DLG4 mice variants will be starting soon at UAB (page 4).

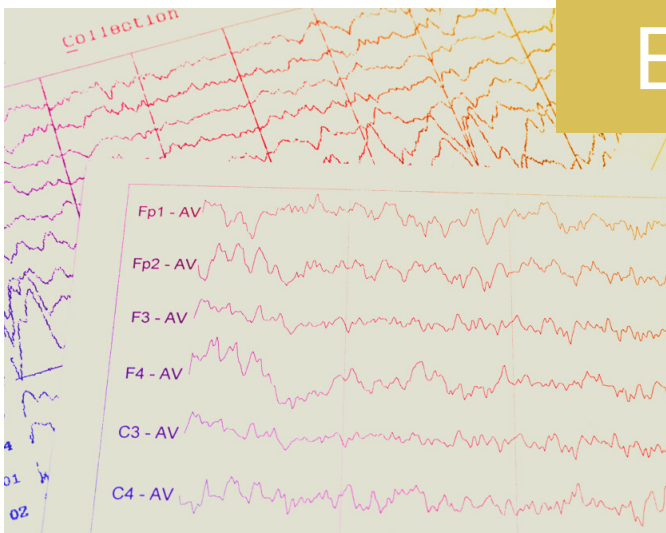
CIITZEN/INVITAE

Ciitizen is another registry platform available to **US patient families**. Ciitizen is a platform where families register and store their medical records. There are also research opportunities where families can choose to share medical information with researchers. Click [here](#) to join the Rare Patient Network.

Join our Rare Patient Network

Now supporting childhood onset of epilepsy and/developmental delay.

[Get started](#)



EEG TRACINGS

Anyone who is participating in the Epilepsy Research Project with the Danish Epilepsy Centre can submit tracings to:

Prof. Guido Rubboli
Adult Department
Danish Epilepsy Center
Kolonivej 2A
4293 Dianalund, Denmark



SOCIAL MEDIA AWARENESS

One of our missions is to raise global awareness about DLG4 and SHINE Syndrome. As part of this mission, our foundation is active on all the major social media platforms. Parents joining forces and helping spread the word is also important and will expedite these awareness efforts. Consider joining these platforms and/or using the following hashtags in posts about your SHINE kiddo: #DLG4 #SHINESyndrome

You can follow our social pages at Twitter, Facebook, Instagram, and LinkedIn. This [linktr.ee](#) will take you to all the pertinent SHINE Syndrome Foundation social media links!



ICD-10 CODE APPLICATION

We submitted an ICD code application officially in late October and have met all subsequent follow-up requests for more information. In early 2023, we should get more information on if our application is chosen to present at the next official ICD-10 meeting in March 2023.

If selected, Dr. Tümer will prepare a slide deck and present it to the CDC. The official diagnosis name in our application is DLG4-related synaptopathy. We will keep you posted about this advocacy effort! Our fingers are crossed our application is successful! An ICD-code would be HUGE for our community.

EPILEPSY PANELS AND GENE REVIEWS

In the fall of 2022, the SHINE Syndrome Foundation requested that the DLG4 gene be added to the three major epilepsy panels. These requests are currently under review.

In December 2022, a team including SHINE MAB members from the University of Copenhagen, CHEO, Lurie Children's, and Cincinnati Children's submitted a pre-approved chapter for DLG4 in Gene Reviews! This is very exciting for our gene.

The Gene Reviews chapter should be available soon and will be published in our family FB group and on all social media platforms.

IPSC LINE DEVELOPMENT

There is a free opportunity to have your child's exact variant included in iPSC line development via Simons Searchlight.

To participate with Simons Searchlight, log into your dashboard. After completing the surveys and medical history interview, request a kit and schedule a blood draw at Quest. This is currently only available to those in the US, though we hope to collect international samples at a future family conference in the summer of 2024. Below is a compilation of iPSC lines and mice models in production or starting soon from all three DLG4 foundations. (Variants are reported in different ways from different institutions).

iPSC lines already obtained from CHEO:

- c.455delG (frameshift)
- c.1054C>T p.R352X (nonsense)

iPSC lines (with parental controls) currently in production via Simons Searchlight:

- Val735Trpfs*12
- Gly79Trpfs*3
- IVS6-2A>G
- Gln401*
- Arg539*
- Glu304fs

iPSC lines currently in production at COMBINEDBrain biorepository:

- c.1961 C>T p.T654I (missense)
- c.1201 C>T p.(Q401*) (nonsense)
- c.1039del p.(E347Rfs*12) (frameshift)

Mice model currently in production: at Cincinnati Children's Hospital:

- c.2203_2207delinsT (frameshift)

Mice model starting soon at the University of Alabama:

- c.1961 C>T p.T654I (missense)

FUNDRAISING EFFORTS



We are pleased to welcome Justin Neduchal to the SHINE Syndrome Board as the Director of Fundraising. Justin is the dad of SHINE star, Jace, and may look familiar to you as his family was featured in the documentary.

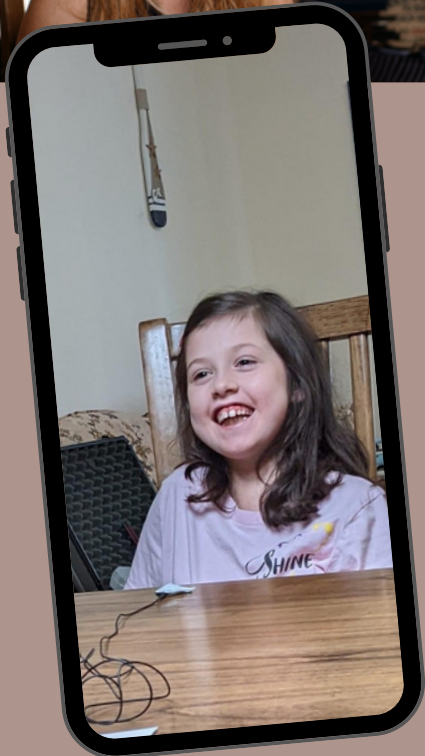
Like all nonprofits, we will always be actively fundraising and the more we work together, the stronger our organization will be. Ways you can help with our fundraising efforts include sharing fundraisers within your community and with friends and family, creating a personal FB fundraiser, and linking with our organization on Amazon Smile.

"Giving is not just about making a donation, it's about making a difference."
- Kathy Calvin

We are also continuing to collect donations in the following ways:

- Via Check
Mail donations to:
SHINE Syndrome Foundation
4906 White Blossom Blvd.
Mason, OH 45040
- Via Venmo
◦ @shinesyndrome
- Via Online Donation
◦ Visit our website for more information:
www.shinesyndrome.org/takeaction

If you have other creative and unique fundraising ideas, send us an email: board@shinesyndrome.org



SHINE SYNDROME DOCUMENTARY

Please check out the SHINE Syndrome documentary. It was filmed by Adam Leigh-Manuell of Blackthorne Studios (Ohio) and Dominic Llanos of Cloud in the Sky studio (Canada). Blackthorne Studios did a fabulous job seamlessly editing this into one cohesive film.

Thank you!

[CHECK IT OUT ON YOUTUBE](#)



SHINE a Light on DLG4

PLEASE SHARE THIS FILM WITH FRIENDS AND FAMILY!

