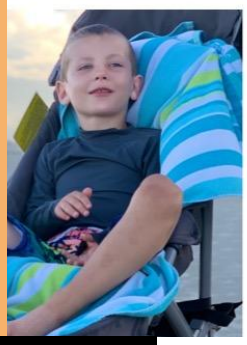
NEWSLETTER SPRING2022










SHINE SYNDROME FOUNDATION, INC

Quarterly Newsletter

SPRING 2022

The Beginnings of our Foundation

A group of like-minded parents, at the urge of medical professionals, began the process of starting a 501c3 nonprofit organization in the fall of 2021. The mission of the SHINE Syndrome Foundation is to improve the quality of life for children and families impacted by SHINE Syndrome through supporting collaborative research among international medical professionals, nurturing a family community, and raising worldwide awareness. We incorporated in the state of Ohio on December 29, 2021, and applied for 501c3 status. We received notice on March 24, 2022, that our application for 501c3 status is approved!

TOP NEWS INSIDE

Simon's Searchlight and CoRDS Registry Information

Why We Are Using Two Registries:
After researching and completing several different registries, our foundation chose to endorse two different registries. We chose Simon's Searchlight since it will compare DLG4-related disorders to almost 200 other neurodevelopmental disorders and we also chose to create a CoRDS registry since it is more personalized to SHINE Syndrome and unique challenges for an individual with a SHINE Syndrome diagnosis. We will roll this out in the second half of 2022.

Simon's Searchlight Information:
Simon's Searchlight offers a set of standardized surveys spanning several different areas of development and milestones. They will also collect DNA from U.S. patients (and international patients when we organize a SHINE family conference) to create stem cell lines made available for researchers.

CoRDS Registry Information:
CoRDS is a registry developed by our foundation and will be instrumental in creating a detailed SHINE phenotype; this could help improve diagnosis rates and will be helpful to providers who may want to compare patient data or effective treatment options. Like Simon's Searchlight, CoRDS can also collect stem cells for research. We hope our families will participate in both. We hope to launch CoRDS this fall!

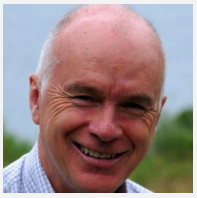
Meet one of our SHINE Stars, Emily!

Emily is a spunky nine-year-old from Columbus, Ohio, USA. On paper, Emily has ADHD, mild developmental disability, developmental coordination disorder, hypotonia, DLG4-related neurodevelopmental disorder, and ESES. It's been a search since birth to pinpoint the diverse mixture of challenges Emily has fiercely endured. We thankfully found the DLG4 community last year and are very thankful for a supportive and informative environment to help us get through this crazy journey. Emily struggles daily with gross and fine motor, sensory, managing her emotions, academics, and life skills. She loves to sing and dance, her smile and laugh light up a room, and she never ever gives up. She also loves to read and watch tv/movies.

Medical Advisory Board

Our SHINE medical board includes specialists in human genetics, neurology, pulmonology, and endocrinology.

Dr. Thomas Dye, *Cincinnati Children's Hospital, Ohio, a child neurologist and sleep medicine specialist focused on better understanding and improving the sleep and circadian health of children, especially those with neurodevelopmental disorders.*



Dr. Alex MacKenzie, *Children's Hospital of Eastern Ontario, attending pediatrician at CHEO and has served as the CEO and Science Director of the CHEO Research Institute as well as Vice President of Research for both CHEO and Canada's federal genomic research funding agency Genome Canada.*

Dr. Carlos Prada, *Lurie Children's Hospital, Chicago, Illinois, is a clinical geneticist very interested in partnering with families to enable better care and research related to rare diseases. He's interested in learning more about the variability of disease manifestations in SHINE Syndrome and how this can help identify future potential therapeutic interventions.*

Dr. Zeynep Tümer, *Copenhagen University Hospital, is a medical doctor and geneticist working with rare neurodevelopmental disorders, including SHINE Syndrome (DLG4-related synaptopathy). In 2021, a large series of individuals with DLG4 variants was published where she was the senior, corresponding author. Since then, Dr. Tümer has been working on different cellular biology projects to understand the underlying pathology of SHINE Syndrome, as this is crucial to paving the way to treatment possibilities.*

Dr. Leanne Ward, *Children's Hospital of Eastern Ontario, a Professor of Pediatrics at the University of Ottawa. Dr. Ward's research program is dedicated to the study of bone development and the diagnosis and treatment of bone disorders in children due to chronic illnesses. Dr. Ward has published extensively in the field of pediatric bone disorders. In 2019, Dr. Ward was named a Fellow of the American Society of Bone and Mineral Research, a lifetime achievement award in recognition of significant contributions to bone and mineral science.*

The Meaning Behind the SHINE Name and Logo

SHINE is an acronym; each letter stands for a common characteristic that comes with a SHINE diagnosis: **Sleep Disorder, Hypotonia, Intellectual Disability, Neurological Disorder,** and **Epilepsy.** The logo was created using the awareness colors for sleep disorders (black), developmental delay (yellow), and epilepsy (purple). The four stars represent the 4 in DLG4 since that is the gene where the mutations occur. The stars also correlate with the word SHINE and how all our children are SHINE-ing lights in our lives. The logo was recently updated by adding the word "syndrome" to increase the visibility that SHINE is a genetic disorder. The SHINE logo was created by Cortny Helmick, a graphic designer and the aunt of one of our SHINE patients. If you haven't purchased any SHINE spirit wear yet, our Bonfire store is open indefinitely. Merchandise ships every three weeks.

A Letter from the President

Three years ago, my son's geneticist encouraged me to start a foundation for SHINE Syndrome. I came up with every excuse as to why I shouldn't do it. It seemed like a lot of work to take on (and it IS a lot of work). But it's important work. Three years ago, we only had 4 families in our family Facebook group. Now, we have almost 60. We are in the baby steps right now, building a foundation that will impact generations to come. My why is my son and all the other SHINE families.

Laura Palmer

How to Support our Cause:

Now that we are officially a 501c3 charitable organization, we can accept donations. There are several ways to make a tax-deductible donation to our Foundation.

1. Send a check:

The SHINE Syndrome Foundation
4906 White Blossom Blvd.
Mason, OH 45040

2. Venmo: @shinesyndrome

Options <u>coming soon</u>:

3. Online on our website, shinesyndrome.org
4. Facebook Fundraiser on our public page

You can also support our cause by purchasing SHINE Syndrome merchandise from our Bonfire store. Merchandise ships every three weeks and a small amount from each purchase comes back to our Foundation.

Our Current SHINE Syndrome Foundation Board of Directors

Our SHINE Syndrome Board is currently comprised of SHINE parents. If you are interested in getting involved or attending one of our monthly board meetings (held the second Sunday of the month at 12:00 EST), email Brian Lareau, Secretary, at blareau@shinesyndrome.org for a meeting invitation.

Laura Palmer, President, *Cincinnati, Ohio, US*
Colleen Lareau, Vice President, *Chicago, Illinois, US*
Nate Palmer, Treasurer, *Cincinnati, Ohio, US*
Brian Lareau, Secretary, *Chicago, Illinois, US*
Marie-France Gervais, Medical Advisory, *Ottawa, Canada*
Brooke Amos, Board Member at Large, *Albertville, Alabama, US*
Courtney Roche, Board Member at Large, Long Island, *New York, US*

Upcoming Events and Campaigns

- **July 17th: SHINE Syndrome Awareness Day**
- **July 1-31: SHINE Fundraising Campaign**
- **Fall 2022: Launch CoRDS registry campaign**

Did You Enjoy the Newsletter?

- Don't miss the next one! Complete this form to receive future digital newsletters via email.
- We'd love to feature your child! If interested, email Laura and/or complete this form to submit photos to use in publications and social media.