NEWSLETTER SUMMER2022









SHINE SYNDROME FOUNDATION, INC

Quarterly Newsletter

SUMMER 2022

What are our Goals?

Now that we are established as a nonprofit, it is time to get to work. 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.**

You can read more about these short- and long-term goals in the infographic found on page three. In the short term, we are doing a SHINE Syndrome Awareness Day on July 17, 2022. Our goal is to raise **$50,000 in the month of July** to fund a postdoctoral researcher in neuropharmacology.

TOP NEWS INSIDE

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RAFFLE ALERT!

SHINE SYNDROME AWARENESS DAY

We are having a VIRTUAL raffle during the month of July using an online site called Give Lively. All funds raised will be used to fund a postdoc in neuropharmacology to partner with our Medical Advisory Board about current FDA-approved drugs that may be beneficial for SHINE Syndrome patients.

Click Here

Ways YOU can get involved!

Our Foundation is looking for a few experts or committed family members to help within the SHINE Syndrome Foundation. Email Laura with interest.

Corporate Fundraising/Grant Writing
Do you have experience with soliciting corporate donations, grant writing, or general fundraising.? We would love to have you join the team!

Family Engagement Specialist
We are looking for a member in our group who is willing to be a friendly, welcoming face when new families join our group. This can be done virtually and may include connecting over FB Messenger, email, and/or phone. Ideally, this person is familiar with the registry process and process for submitting family photos for publicity.

U.S.-based Accountant with Non-profit Experience
At year-end, we will need assistance with filing taxes. Our Foundation is U.S.-based, specifically incorporated in Ohio. Please reach out to treasurer, Nate Palmer, if you can help in this capacity.

Epileptologist with ESES experience
The last role we need to fill on our Medical Advisory Board is an epileptologist. The criterion we are interested in includes:

- currently treating 1+ patient with SHINE who has epilepsy
- experience and knowledge with epileptic encephalopathy and ESES
- speaks English

Email Mara with any recommendations!

Meet one of our SHINE Stars, Izac!

Izac is almost 10 years old. He is from Shuswap, BC Canada. We are fortunate enough to have worked with some amazing professionals to pinpoint what Izac's struggles are. Izac struggles with: Sleep disorder, Hypotonia, Intellectual Disability, Neurological disorder, and epilepsy. Izac has a favorite teddy named "Blue Dog": he is a world traveler, quading machine, sleep companion, and has been patched from head to toe. Blue Dog is a huge part of our family. Izac is there when you need help and he is the first one to know when you are sad. He is definitely the comedian in the family–he makes us laugh a lot. To all the families that share similarities we are beside you, we hear you, and we know you are out there.

SHINE SYNDROME FOUNDATION

FUTURE PLANS

SIMONS SEARCHLIGHT

We would love to see at least 75% of our families complete the registry process for Simon's Searchlight.

CORDS

CoRDS will be our second registry and will be more specific to SHINE Syndrome than Simons, which compares many neurodevelopmental disorders.

IMPROVE DIAGNOSIS

We are currently working on getting the DLG4 gene added to the most-used epilepsy panels as a goal to improve diagnosis rates.

FAMILY CONFERENCE

Our early goals include a family conference in 2023 to give families an opportunity to meet in person and learn more about this disorder from experts.

RESEARCH

Currently, Dr. Tümer is writing a proposal to do an intensive study into the effects of PSD-95, particularly in missense variants and to study the neurodegenerative prospects of this diagosis.

NORD

Our Foundation is currently in the application stage for platinum membership with NORD (The National Organization for Rare Disorders).

SHINE ICD CODE

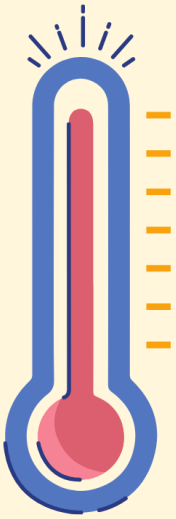
For years, many of us went undiagnosed. We are working on an application to grant an official ICD code for this disorder.

FAMILY ENGAGEMENT

We would LOVE to see more families become involved. See the opportunities section of the newsletter for specific needs or reach out if you have another way to help!

For more details, you visit www.shinesyndrome.org

HERE'S HOW YOU CAN HELP



Current Fundraising Project

Postdoctoral Researcher:
Hire a dedicated postdoctoral researcher specializing in neuropharmacology to work with our Medical Advisory Board to find currently available drugs that may increase PSD-95 in patients.
GOAL: $50,000

SHINE

AT A GLANCE

www.simonssearchlight.org/research/whatwestudy/dlg4/

98% — Percentage of SHINE Syndrome patients diagnosed with an intellectual disability.

Percentage of patients who showed developmental delays in early childhood. — 84%

53% — Percentage of SHINE patients who experience epilepsy. Seizure types can range and several have a developmental epileptic encephalopathy.

Percentage of patients who have a diagnosis of autism spectrum disorder. — 55%

A Letter from the Vice President

From the moment the doctor said we were having twins life has never been the same. Identical twins carry their own relatively "rare" title, so having twins with a rare genetic condition felt like an even bigger shock. Luckily with the support of our families, school and therapy teams and our SHINE community we've managed to watch our boys excel. It's not always a graceful or scenic ride but we wouldn't have traveled this far without each person encouraging us. We thank you all.

Colleen Lareau

How to Support our Cause:

There are several ways to make a tax-deductible (US only) donation to our Foundation.

1. Send a check:

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

2. Venmo: @shinesyndrome
3. Donate on our website, shinesyndrome.org/take action
4. Facebook Fundraiser on our public FB page
5. Purchasing SHINE merchandise from our Bonfire store.
6. Purchasing Raffle tickets via GiveLively for our July fundraiser
7. Sharing our fundraisers with friends and family!

Meet the Newest Addition to our Medical Advisory Board

Alexandre White-Brown
Children's Hospital of Eastern Ontario

Alexandre White-Brown is an MSc Genetic Counseling candidate currently working as a Clinical Neurogenetics Coordinator at the Children's Hospital of Eastern Ontario (CHEO). He works directly with the Canadian Care4Rare Research Team focusing on the diagnosis and treatment of rare genetic disorders. Alexandre also has over 4 years of experience providing respite care for a child with SHINE Syndrome.

Upcoming Events and Campaigns

- **July 17th: SHINE Syndrome Awareness Day**
- **July 1-18th: Virtual Raffle**
- **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.