# STXBP1 disorders
Science + Love = Cure

## Welcome to the November 2023 issue of the STXBP1 Foundation Newsletter


### FDA EL-PFDD Comments Still Open

Our recent record breaking EL-PFDD meeting with the FDA was a reflection of community, care, and compassion and it highlighted the urgent need for treatments. It helped paint a sometimes beautiful, and sometimes painful picture of what it is like to live with STXBP1-related disorders. This is a picture that the FDA will look at when considering approvals for future treatments. The STXBP1 Foundation would like to thank everyone who joined the meeting, participated, and has commented on our website thus far.

**It's not too late to comment! You have until November 19 to comment on our website**, here.

---

GIVING TUESDAY

## Giving Tuesday Is Coming Up

We would like to take a moment to thank each and every person who is reading this newsletter for giving us your valuable time and for all of the love and support you've shown over the years. We are very grateful to all of our partners and donors who are helping us progress toward an improved quality of life for those with STXBP1-related disorders. In pursuing our mission, the support we have received from partners and the community have made all the difference in the world. The generous donations we have received throughout the years have provided critical resources and momentum for our important cause.

**Please consider donating here to help us find a cure for epileptic encephalopathy and please consider sharing our social media posts to ask others in your community if they'd be willing to help too - spread the word!**

---

AES AMERICAN EPILEPSY SOCIETY

### STXBP1 Foundation at American Epilepsy Society Conference

We will be at the American Epilepsy Society (AES) conference this year to support our mission by raising awareness and forming partnerships. This important event brings healthcare providers, scientists, researchers, and advocates together to collaborate on how to improve outcomes and quality of life for people with epilepsy. We will be at AES in the Exhibit Hall's Epilepsy Resource Area excited and ready to share our mission and message.

**Find out more about the conference here. If you plan to attend the event, come visit us in booth N750, or if you are in Orlando area and would like to meet other STXBP1 parents email us here.**

---

STARR The STXBP1 Natural History Study

### STARR Clinic Updates

The STARR Natural History Study is alive and well with 45 participants seen in the clinics and another 20 scheduled to be seen by the end of the year! We now have the following sites accepting patients: The Children's Hospital of Philadelphia, Children's Hospital of Colorado, Texas Children's Hospital, Weill Cornell Medicine. All 4 sites are currently enrolling participants and working to schedule all participants to be seen at the site. Make sure that you make plans to participate at your nearest location if at all possible so we can continue our important efforts to secure clinical trial readiness!

**Visit our website here for more information on how to get started with the STARR Natural History Study.**

---

## It's Epilepsy Awareness Month


November is Epilepsy Awareness Month! With 85% of those with STXBP1-related disorders experiencing seizures, many within our community are profoundly impacted by them.

We would like to take a moment to recognize the strength and resilience of all about patience for enduring the great difficulties that seizures can bring. One of our board members wrote a post about his first experience with seizures. He begins with the phrase "holding your own child as she has a seizure is like holding your heart as it beats outside of your body."

**Read more of our blog post and share your own experiences with seizure here.**

---

CLARA INSPIRED

### Clara Inspired has Funded a New Nonsense Mouse Model

STXBP1 Foundation is working to create a specialized nonsense mouse model for STXBP1. This model will mimic the effects of nonsense mutations, providing invaluable insights into the condition and potential treatment avenues. A nonsense mutation is a change in DNA that causes a protein to terminate or end its translation earlier than expected. Some in our community have nonsense mutations. The development of this mouse model, a critical investment in advancing STXBP1 research, was generously donated by Clara Inspired. Thank you Jackson Labs and Clara Inspired for helping ensure this vital resource will be accessible to researchers!

**Read our blog post to learn more about this exciting development here.**

A nonsense mutation, or its synonym, a stop mutation, is **a change in DNA that causes a protein to terminate or end its translation earlier than expected**. This is a common form of mutation in humans and in other animals that causes a shortened or nonfunctional protein to be expressed.

---

### STXBP1 Summit+ 2024


We're excited to be headed back to the Philly area for the 2024 STXBP1 Summit+ Researcher & Family Meetings!

**Mark Your Calendars**
Researcher Meeting - July 18-19
Family & Researcher Dinner - July 19
Family Meeting & Support Sessions - July 20-21

---

Read our past issues here. We'll see you next month.



**Our mailing address is:**
STXBP1 Foundation
P.O. Box 1148
Holly Springs, NC 27540
USA