STXBP1 Disorders

Leading the Charge For a Cure

Welcome to the November issue of the STXBP1 Newsletter.

First off, it was a very memorable STXBP1 Awareness Month in September.... ***Because Of You***!

Highlights included:

- **4th Annual Move to Cure** event which included more than 250 people across 4+ countries and 30+ states in the USA
- **Researcher Update** Webinar series
- **Did You Know cards** - this year in English, Dutch, German and Spanish!

The goals of STXBP1 Awareness month was to increase awareness of STXBP1, raise funds for research and have some fun too. We were able to raise $65,000 through Move to Cure!

If you missed any of the fun, check out our blog post here.

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## Researcher Webinar Series - Recordings Available!

- **Ingo Helbig & Sarah McKeown:** STXBP1: An Update
- **Matthijs Verhage & team:** Designing a STXBP1 Natural History Study
- **Ben Prosser & Ganna Balagura:** ASOs and Genetic Therapies
- **Wendy Chung:** Simons Searchlight and STXBP1 Registry Update
- **Jacqueline Burre & team:** 282 STXBP1 Patient Review & Small Molecule Therapies
- **Zach Grinspan:** 4-Phenylbutyrate Clinical Trial Pilot
- **Mingshan Xue:** How Different Neurons in the Brain Contribute to STXBP1 Encephalopathy

The full set of webinar recordings are now available here.

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## Save the Date… Giving Tuesday is December 1st!


Giving Tuesday is an international day of giving that was started in 2012, and has turned into a global movement.

Our goal is to raise $15,000 on Giving Tuesday, and an additional $10,000 by the end of the year. With your help we know that we can meet our goals!

Follow us on social media on December 1, where we will provide opportunities throughout the day to make an impact by donating in honor of children with an STXBP1 disorder.

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## Rare Bear Drive for STXBP1 Patients

If you don't yet have a RARE Bear for your STX'er, register for a one of a kind RARE Bear here: https://www.rarescience.org/stxbp1/

The campaign will be open until December 15!

Learn more about the RARE Science RARE Bear Program here.

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## Survey on Quality of Life & Non-Seizure Symptoms: Share Your Experience!

CHOP is running a survey on non-seizure endpoints and patient quality of life for patients with epilepsy and/or neurodevelopmental disorders. We want STXBP1 experiences to be represented in this survey!

The University of Pennsylvania and Children's Hospital of Philadelphia researchers recognize a need for proper diagnosis and treatment of symptoms associated with neurodevelopmental disorders as well as the associated effects on patient quality-of-life. If you agree to take part in this study, they will ask you to answer questions about your demographics as well as your child's medical history and quality-of-life.

The questionnaire will only take 20 minutes.

Participate in the survey here: https://redcap.chop.edu/surveys/?s=73ELXYD7H8

The survey will be open until December 15!

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## New Board Member: Jackie Steinberg

We are delighted to announce that Jackie Steinberg, BSN, MPH, CIC has joined the STXBP1 Foundation Board of Directors. Jackie has a 2 year old STX'er, and she is an Infection Control Nurse at Johns Hopkins Bayview Medical Center. Jackie has represented the foundation & STXBP1 at the American Epilepsy Society Conference as well as Rare Disease Day at the NIH last February. Jackie is also an active participant in the Million Dollar Bike Ride as part of Lulu's Crew Team STXBP1, helping to raise funds for STXBP1 research.

Please join us in welcoming Jackie and we thank her for taking on this volunteer role.

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## STXBP1 Foundation is Rare Champion of Hope Nominee 2020

Global Genes honors individuals and foundations who are making exceptional efforts when it comes to rare disease advocacy and change through its Rare Champion of Hope Celebration. This year, the STXBP1 Foundation was nominated as a Rare Champion of Hope Nominee 2020. Learn more here.

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## New STXBP1 Regional Groups

Our new regional groups launched this Spring; get involved! We want to bring STXBP1

their regions. We know we can do this online as well as in person. The groups are starting to take off, from planning future meet ups and participating in virtual fundraisers, to supporting and caring for their local STXBP1 families in the hospital.

Currently we have 8 regions around the US with 5 or more families that have formed regional groups. Additional regions are looking for a few more families, or parents to step up to lead the region. If you are interested in helping form a new regional group or joining a current regional group, contact community@stxbp1disorders.org.

- Northeast (NY, NJ, MA, CT)
- Mid Atlantic (MD, DE, VA, Eastern PA, Eastern NC)
- South East Region (TN, AL, GA)
- Florida
- Ohio River Region (OH, WV, Western PA)
- Michigan
- Texas
- California

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## Fundraising for STXBP1


The global pandemic has created many challenges for us and other nonprofits who rely upon fundraising to fund our mission. Through all of this our mission has not changed, we are still committed to finding a cure for STXBP1 disorders. Please consider donating or creating a fundraiser of your own during our Giving Tuesday campaign on 12/1, or a year end donation to help us meet our fundraising goal of $25,000.

We are a 501c(3) non profit, and all donations are tax deductible.

Also check out the STXBP1 Foundation Bonfire store! Proceeds go to STXBP1 research and clinical trial readiness.

the sale will come directly to us.

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