Welcome to the August issue of the STXBP1 Newsletter!

September is **STXBP1 Awareness Month**! This is our second year of dedicating the entire month of September to STXBP1 with ongoing communications and activities.

Why September? September is the 9th month and the STXBP1 gene is on the 9th chromosome.

Join in all month long and we'll work together to create more awareness for this rare genetic disorder, keep up with the very latest research, and bring the community together through our annual Move to Cure event … virtually!

4th Annual Move to Cure STXBP1 Disorders

The 4th Annual Move To Cure STXBP1 Disorders event will take place at the end of STXBP1 Awareness Month, 26 - 30 September. We are challenging the community to move 9 miles - whether that's among 9 friends and everyone walks one mile, or one partner runs 3 miles 3 times or you hike a 9 mile trail! However you can get 9 miles! 9 miles because STXBP1 is on the 9th chromosome!

Last year, we had 250+ people around the world participate and we can't wait to see how the community shows up this year. If you missed last year's event, here's a video of highlights from last year!

We appreciate that this year's event will look different. If your state or city will allow you to

Let's raise awareness for STXBP1, raise funds for research and have some fun while we're at it!

Sign up here: Move to Cure Virtual Challenge sign up

Buy custom shirts from Bonfire here: Team shirt

Questions? Contact Allison Michels.

STXBP1 Awareness Month: Webinar Series

Get your STXBP1 Research Updates Here! This September, we will hold a series of virtual updates from researchers so our community has the latest research information.

We'll hear from these confirmed speakers:

- **Matthijs Verhage & team:** Designing a STXBP1 Natural History Study and Verhage Lab Updates
- **Ben Prosser:** ASOs and Genetic Therapies as a Therapeutic Approach for STXBP1 Encephalopathy
- **Wendy Chung:** Simons Searchlight and STXBP1 Registry Update
- **Jacqueline Burre & team:** 282 STXBP1 Patient Review and Small Molecule Therapy Approaches
- **Zach Grinspan:** 4-Phenylbutyrate Clinical Trial Pilot
- **Mingshan Xue:** How Different Neurons in the Brain Contribute to STXBP1 Encephalopathy

Learn more and sign up for these webinars here.

$143K Raised for STXBP1 at Million Dollar Bike Ride

The Million Dollar Bike Ride took place on Saturday, June 13. Lulu's Crew Team STXBP1

With these funds, $143K in seed grant funding will be available for STXBP1 research through University of Pennsylvania's Orphan Drug Center! Letters of intent are due September 18; make sure your favorite researcher is aware of this opportunity! More information and the request for applications are posted on the ODC site.

Researcher Spotlight: Michael Boland

This issue's Researcher Spotlight features Dr. Michael Boland. Michael is Assistant Professor in the Department of Neurology and Director of Cellular Models of Disease at the Institute for Genomic Medicine at the Columbia University Medical Center. His work focuses on pediatric epileptic encephalopathy, including those caused by STXBP1 mutations, and monogenic autism spectrum disorders.

Michael made STXBP1 a research focus when his infant son was diagnosed with a de novo loss of function mutation in STXBP1.

He and collaborator Wayne Frankel were just awarded a multi-year grant by SFARI to develop and test multiple genetic approaches for the ability to correct phenotypes in cellular and mouse models of STXBP1.

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100 Ultrasong Challenge Fundraiser

Father of STX'er Clara, Casey Baum has been inspired again this year for a marathon. This time, he'll be performing 100 songs over 8 hours. On August 22, listen in and donate to Clara Inspired for rare genetic disease awareness and research!

New STXBP1 Regional Groups

Our new regional groups launched this Spring; get involved! We want to bring STXBP1 families together to build a network of regional support. We are empowering local families to connect with each other, advocate, and build relationships with other STXBP1 families in 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

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. All of our in-person fundraisers have been postponed this year, so we are having to get super creative with our fundraising efforts. Watch our Facebook page and website for upcoming virtual fundraisers.

Do you have a fun idea for a virtual or socially distanced fundraiser? We would love to hear it. Please email melissa.hioco@stxbp1disorders.org with your ideas!

Also the new STXBP1 Foundation Bonfire store has launched! Proceeds go to STXBP1 research.

Registry

he STXBP1 registry is an important method for increasing what we know about STXBP1, and for developing future treatments and therapies. Researchers need an adequate pool of participants to make their studies statistically significant. And, we streamline research efforts by hosting our information in one central location. See how easy it is to join our registry, hosted with Simons Searchlight, and read the most common FAQs here.

Also don't miss Wendy Chung's update on Simons Searchlight and the STXBP1 Registry on September 19!

Upcoming Research & Advocacy Conferences

The Epilepsy Foundation Pipeline Conference (8/26-27) and the Global Genes RARE Patient Advocacy (un)Summit (9/14-25) are both virtual this year. The Global Genes (un)Summit is free to patient advocates this year! The STXBP1 Foundation attend both of these meetings. If you are interested in attending, please visit their registration sites or contact us.

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