STXBP1 Disorders

Leading the Charge For a Cure

Welcome to the February issue of the STXBP1 Newsletter.

Despite the global challenges due to the coronavirus pandemic, the perseverance and passion of our community yielded multiple successes and highlights for the foundation and our community. Together we continue to advance research, connect families, and expand awareness of STXBP1 disorders. 2020 highlights include our Research Webinar series, the 4th Annual STXBP1 Move to Cure with more than 250 participants, and raising over $300,000 to fund STXBP1 research and awareness initiatives.

For a 2020 recap, check out our blog post here.

4-Phenylbutyrate Clinical Trial Is Recruiting

The clinical trial pilot for 4-phenylbutyrate is starting recruitment! This is the first clinical trial with a therapy targeting the STXBP1 protein.

We are thankful to Jacqueline Burre, PhD and Zach Grinspan, MD, MS and their teams at Weill Cornell, for their work that started in the lab to identify 4-phenylbutyrate through to this clinical trial.

For information on 4-phenylbutyrate and the trial, including inclusion criteria and how to contact the study team, check out our blog post on the clinical trial.

This clinical trial has been funded by grants from the ODC's Million Dollar Bike Ride - Lulu's Crew Team STXBP1, and Clara Inspired. In addition, the STXBP1 Foundation and Clara Inspired are offering a Patient Assistance Fund to cover unreimbursed medical and travel expenses up to a set amount per enrolled patient.

Double Your Donation for Rare Disease Day 2/28!

February 28 is World Rare Disease Day. STXBP1 Disorders is one of the 7,000+ Rare Diseases, and our families are among the 300 million affected by a rare disease. 95% of these rare diseases lack an approved treatment. We want to change this for STXBP1 Disorders. We are making great progress, and with your help we can continue our mission to accelerate treatments and hopefully a cure.

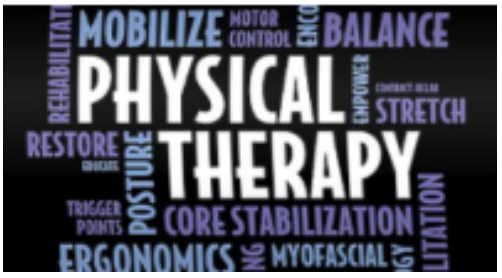
The STXBP1 Foundation Board members are matching donations up to $6,000 from now through Rare Disease Day on February 28…right out of our own pockets!

To give, simply text the code 147231 to 1-855-575-7888 or go to our website at https://www.stxbp1disorders.org/donate-index-impact to make a tax deductible donation. All donations directly fund and support research efforts. Also, donations on our website can be made in honor or memory of a loved one.

STXBP1 at NIH Rare Disease Day

Parent and Foundation Board Member Jackie Steinberg will be representing STXBP1 at the NIH Rare Disease Day on 1 March. The event is virtual this year so you can join from anywhere. Want to join and represent STXBP1? Contact us.

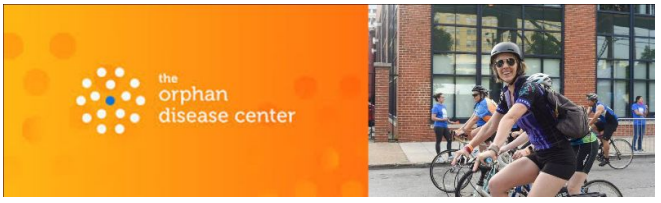
Webinar 3/16: Physical Therapy for Children with STXBP1



Dr. Samuel Pierce, PhD, NCS, from Children's Hospital of Philadelphia will present a webinar on interventions and equipment typically used with children with STXBP1 Disorders. He will also discuss the role of physical therapists as part of the treatment team for STXBP1 patients.

The webinar will be held on 16 March at 12 pm ET. Register here.

ODC's Million Dollar Bike Ride Grant Awards



The Orphan Disease Center (ODC) has announced the 2020 Million Dollar Bike Ride (MDBR) research grant recipients! Two $71,658 grants were awarded to STXBP1 research projects.

One grant went to Jimmy L. Holder, Jr, MD, PhD at Baylor College of Medicine / Texas Children's Hospital. The other grant was awarded to Christopher Makinson, PhD at Columbia University. More information on the projects is here.

We are excited about the potential of these projects to further our understanding of STXBP1 and enable development of therapies for STXBP1, and appreciate these committed researchers.

Thanks to Lulu's Crew Team STXBP1!

Million Dollar Bike Ride 2021

And that brings us to 2021's Million Dollar Bike Ride! Lulu's Crew Team STXBP1 is riding again this year. The MDBR will be a virtual event on June 12th -- so you can join from anywhere! On your road bike, mountain bike, Peloton, or even your tricycle! We are looking for riders, and supporters. Consider joining the team or cheer us on virtually!

In 2020, with $143K total money raised, STXBP1 had the largest grant funding of all the MDBR disease teams. For 2021, let's try to beat that to fuel more research in STXBP1!

To register please complete the online registration: https://www.milliondollarbikeride.org/, then reach out to info@stxbp1disorders.org to set up your fundraising page, or for help to update your fundraising page from last year!

Clinical Trial Readiness

With the 4-phenylbutyrate clinical trial, the ASO program at UPenn and other projects, we are expanding from basic research to therapy development. To learn more about the types of projects and steps from basic research to approved therapies, check out our new Roadmap to Cure infographic. Watch for content and webinars on clinical trial readiness

Survey on Transitions to Adult Care: Share Your Perspective!

The International League Against Epilepsy (ILAE) is sponsoring a research study on the "Perception of transition from pediatric to adult healthcare system in patients with epilepsy". This is an initiative of the ILAE Transition in Care from Childhood to Adult Task Force, and the study is being led by Dr. Danielle Andrade at the Toronto Western Hospital.

Transition is the planned process of moving patients with child-onset epilepsy to adult care. Transition usually starts several years before the patient becomes adult and ends when the patient stops seeing a child neurologist. For background on the study, watch this video from Dr. Andrade.

Families of patients 12 and above, including patients who have already completed transition to adult care, can participate. We want STXBP1 experiences to be represented in this study.

To learn more or participate in the study, please contact Quratulain Zulfiqar Ali, MD at Quratulain.zulfiqar_ali@uhn.ca.

The study will be open until June 30, 2021.

New Seizure Action Plan Resource

The Seizure Action Plan (SAP) Coalition just launched to educate people with epilepsy, their caregivers and healthcare professionals about seizure emergency rescue protocols and the importance of personalized seizure action plans.

A seizure action plan contains tailored guidelines on how to respond during a seizure, based on the patient's medical history.

The Seizure Action Plan Coalition website provides resources to create a personalized Seizure Action Plan - in English, Spanish, Chinese and other languages.

Join an STXBP1 Regional Group!

Our regional groups launched in 2020 and are growing. 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. Please consider donating in recognition of Rare Disease Day, on 28 February.

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.

Finally when shopping on Amazon, don't forget to use smile.amazon.com and select the STXBP1 Foundation as your nonprofit. Every time you shop, a portion of the proceeds of the sale will come directly to us.

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