Welcome to the inaugural issue of the STXBP1 Newsletter!

The STXBP1 Foundation is dedicated to raising awareness and finding a cure for STXBP1 disorders. We are excited about the progress we are making as a community, yet we have so much more to do for our STXBP1 patients and families. We look forward to continue working with our community, as together we *Lead the Charge for a Cure*.

2019 Highlights

2019 was an incredible year for the foundation and our community. Together we are advancing research, connecting families, and expanding awareness of STXBP1 disorder. Highlights include:

- Our first STXBP1 Investigator and Family Meeting with over 40 researchers and clinicians as well as 60 families represented
- STXBP1 registry grew to 135 registered families with 63 medical history interviews completed
- First research funding from the foundation, $75K to Ben Prosser's lab at University of Pennsylvania for their promising microRNA therapy work
- $139K raised for STXBP1 research through the Million Dollar Bike Ride put on by the Orphan Disease Center (thanks Lulu's Crew Team STXBP1!)
- The Annual STXBP1 5K expanded to 27 cities and over 230 participants

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New STXBP1 Foundation

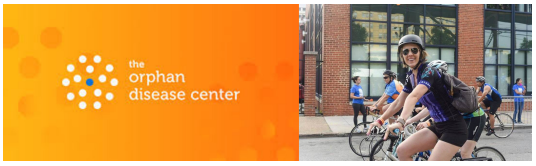
We just launched a new website with an updated look and feel, and expanded content including information on STXBP1 researchers and projects. We welcome your feedback on the new site.

New Research Study from Baylor / Texas Children's Hospital

Baylor College of Medicine in affiliation with Texas Children's Hospital is conducting two studies: one to understand the frequency and severity of sensory processing symptoms in individuals with syndromic autism, and the second on sleep patterns in individuals with neurodevelopmental or neuropsychiatric disorders. The format is a 15-minute survey, either in person or via phone. To participate: read more information here, or contact Maria McCormack, Research Coordinator at +1.832.824.8775 or maria.mccormack@bcm.edu.

Information on other research studies, including an STXBP1 study for patients 18 and older at Children's Hospital of Philadelphia and multiple European centres, available on our website. We encourage you to consider participating in research. Broad patient participation will help us better understand STXBP1 and its symptoms, which will accelerate therapy development.

ODC's Million Dollar Bike Ride Grant Awards



The Orphan Disease Center (ODC) has announced the 2019 Million Dollar Bike Ride (MDBR) research grant recipients! Two $69K grants were awarded to STXBP1 research projects. One grant went to Co-PIs Dr. Matthijs Verhage at Vrije Universiteit Amsterdam and Dr. L. Neukomm at University of Lausanne. The other grant was awarded to Dr. Zachary Grinspan at Weill Medical College of Cornell University for the 4-phenylbutyrate clinical trial pilot. More information on the projects is here. We are excited about the potential of these projects to further identification and development of therapies for STXBP1, and appreciate these committed researchers. Thanks to Lulu's Crew Team STXBP1 for being the driving force behind the inaugural MDBR for STXBP1 research.

Million Dollar Bike Ride 2020

And that brings us to 2020's Million Dollar Bike Ride! Lulu's Crew Team STXBP1 has already been accepted as a Disease Team by the Orphan Disease Center for the Million Dollar Bike Ride 2020. The ride will take place on June 13th in Philadelphia, PA. We are looking for riders -- including virtual riders -- and supporters. Consider joining the team or be there to cheer us on if you can!

In 2019, with $139K total money raised, STXBP1 had the largest grant funding of all the MDBR disease teams. For 2020, let's try to beat that to fuel more research in STXBP1!

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

1st STXBP1 Foundation Funding to University of Pennsylvania

Last November, the STXBP1 Foundation made its first-ever research funding award, $75,000 to the University of Pennsylvania, Perelman School of Medicine. Principal Investigator Ben Prosser is pursuing a promising genetic therapy approach that targets microRNAs, or miRNA. miRNA-based therapies are quickly emerging as a new type of therapy which allows targeting of specific genes. In the case of STXBP1, the goal is to increase STXBP1 in neurons using an miRNA-based therapy. Ben is a fellow STXBP1 parent. His original research was focused on cardiac disease, but when his daughter was diagnosed, the passion and love that many of our parents share motivated him to switch some of his research focus to the brain. Find out more here in the poster from our STXBP1 Investigator and Family Meeting held June 2019.

Don't Miss the STXBP1 Rare Disease Day Bonfire Shirts!

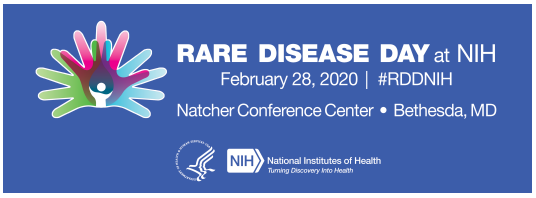
During the month of February we are celebrating Rare Disease Day with an exciting challenge…look for our email and posts on Facebook for all of the details.

And, you can still order STXBP1 Rare shirts through the end of February! Two different STXBP1 Rare shirt designs are available. Each shirt has its own Bonfire order page.

https://www.bonfire.com/stxbp1-i-love-someone-rare/

https://www.bonfire.com/unique-special-rare-stxbp1/

STXBP1 at NIH Rare Disease Day



Parents Jackie Steinberg and Stephanie Wolf will be representing STXBP1 at the NIH Rare Disease Day on 28 February, in Bethesda, Maryland. In the DC-area and want to join them? Contact us.

Fundraising for STXBP1

Interested in holding a fundraising event in your area? If you have an idea or are looking for one, please contact Melissa Hioco, who leads our Development Committee.

Join the STXBP1 Registry

The 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.

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