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Welcome to the May issue of the STXBP1 Newsletter.

Million Dollar Bike Ride, FDA Grant, STXBP1 Mom Heather's 48 Mile Run, New IPSC Lines, Ciitizen Natural History, cryptocurrency *and more*.

Let's get on with the updates!

FDA Grant for ORCA Communication Scale



Duke University School of Medicine and COMBINEDBrain have been awarded a multi-year grant by the FDA to evaluate and expand the Observer-Reported Communication Ability (ORCA) scale for multiple neurodevelopmental disorders.

STXBP1 is one of the first members of COMBINEDBrain to use the ORCA scale. Communication challenges have been consistently identified by our families as a top concern. The ORCA scale would potentially allow us to more accurately describe the baseline communication skills of our children, and ultimately allow us to measure potential changes in communication during clinical trials.

The ORCA scale is being used on an exploratory basis in the STXBP1 4-phenylbutyrate clinical trial now underway at Weill-Cornell. Now, this grant will allow for the ORCA scale to be assessed and validated for multiple conditions including STXBP1. Learn more and read the [press release here](#).

Time for the Million

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The Million Dollar Bike Ride will be a **virtual event on Saturday, June 12**. 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! 100% funds go directly to research. And, Lulu's Crew Team STXBP1 will receive matching funds from the University of Pennsylvania Orphan Disease Center. **Our goal is to fund two seed grants**; your contributions are important to this goal!

Last year, STXBP1 grants from the MDBR and Orphan Disease Center [funded important research projects](#) with Christopher Makinson, PhD at Columbia University, and Jimmy L. Holder, Jr, MD, PhD at Baylor / Texas Children's Hospital.

Support one of these STXBP1 family riders:

- Erin Prosser (Lucy's mom): <http://givingpages.upenn.edu/lulucrewErin>
- Erin Moyer (Benjamin's mom): <http://givingpages.upenn.edu/lulucrewErinM>
- Cassie (Nate's mom): <http://givingpages.upenn.edu/lulucrewCassie>
- Jackie Steinberg (Alex's mom): <http://givingpages.upenn.edu/lulucrewJackie>
- Charlene Son Rigby (Juno's mom): <http://givingpages.upenn.edu/lulucrewCharleneR>

To ride with Lulu's Crew Team STXBP1 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!

Camden's Cure GoFundMe & Mom Heather's 48 Mile Running Challenge



STXBP1 parents, Heather and Matt Jones, have launched a GoFundMe campaign to raise money for research for STXBP1. They have almost reached half of their goal!

Follow along on Instagram [@cureSTXBP1](#) as Heather completes a **48 mile running challenge** on May 29 & 30 to raise funds and awareness!

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100% of proceeds go to research.

Citizen Natural History Study for STXBP1 Opens



The STXBP1 Foundation has launched a [Natural History Study for STXBP1 with Citizen](#). Natural history studies help us to increase what we know about STXBP1 patients, and accelerate development of therapies. The Citizen platform extracts and structures medical record information across multiple hospitals and providers where a participant has been seen. Parents also have access to their child's records in a secure patient portal.

The first cohort of 50 participants from the US filled in April in 4 days! Enrollment for the second 50 participants will start in mid-July. In this second cohort, we will expand to include individuals in countries with medical records in English, with additional languages supported in the future.

[Join the Waitlist](#) to be notified when enrollment of the second Citizen cohort opens.

The first set of de-identified data will be accessible to researchers in September 2021. For more information or to request access, please reach out to research@stxbp1disorders.org.

New iPSCs at Simons Foundation



Induced Pluripotent Stem Cells are an important model in STXBP1 research. We are excited to announce that the Simons Foundation is generating 15 iPSC lines from STXBP1 participants in Simons Searchlight. The first six lines will be available in June; nine more will be available in the fall. More information on the program and specific iPSC lines is [here](#). Researchers can request access through [SFARI Base](#).

We are very appreciative of our long-standing partnership with Simons Searchlight!

Calling all Grandparents

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Grandparents, join the Grands Society!

STXBP1 Grandmother, Helen Rigby, is forming a grandparents group within our STXBP1 community, so grandparents can connect, stay informed, and support STXBP1.

Read her [blog post](#), and reach out to grands@stxbp1disorders.org to join.

Save the Date: Family Meeting Sept 18 & 19



Save the date for our 2021 STXBP1 Family Meeting! This year's meeting will be held virtually on the weekend of September 18 & 19.

Get ready for research and clinical updates, informative and interactive sessions, and time to socialize and catch up with your STXBP1 Family. More details coming soon!

Have a topic idea you would like to see included? Reach out with ideas at info@stxbp1disorders.org.

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](#).

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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 Zulfikar Ali, MD at Quratulain.zulfikar_ali@uhn.ca.

The study will be open until June 30, 2021.

Recap of Rare Bear Campaign for STXBP1



55 families received Rare Bears from our Fall/Winter 2020 Drive, in partnership with Rare Science!

Miss out on getting a bear for your STX'er? We will work with Rare Science on 2021 Rare Bear Drive for STXBP1 in September, during STXBP1 Awareness Month. Thank you, Rare Science!

Join an STXBP1 Regional Group!



Our regional groups have been growing despite the pandemic! Get involved to build a network of STXBP1 families for regional support. The groups are starting to take off. Although the pandemic prevented in person get togethers, it did not deter those groups from rising to the occasion when one of their members needed extra support and love.

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)

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- [California](#)

STXBP1 Now Accepts Donations of Stocks and Cryptocurrency



NEW for 2021! The STXBP1 Foundation is now able to accept gifts in the form of stocks, bonds, mutual funds and cryptocurrency! Please reach out to heather.jones@stxbp1disorders.org to find out more information and how this type of giving could benefit YOU!

Have other ways you would like to give in order to support our mission and our families? Please let us know!

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

Also don't forget these other ways to donate and participate:

- Check out the [STXBP1 Foundation Bonfire store!](#) Proceeds go to STXBP1 research and clinical trial readiness.
- When shopping on Amazon, 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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