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Welcome to the May issue of the STXBP1 Newsletter!

Coronavirus has thrown many of our lives into chaos and uncertainty. During this challenging time, the STXBP1 Foundation is maintaining focus on supporting our community, as well as accelerating therapies and finding a cure for STXBP1 disorders.

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## New STXBP1 Regional Groups



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](mailto: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

## Million Dollar Bike Ride 2020 - NOW VIRTUAL!



The Million Dollar Bike Ride takes place on Saturday, June 13, and is now *virtual*! There is still time to ride with or support Lulu's Crew Team STXBP1. Since this year's event is virtual, you can ride outside or on a trainer, run, walk, scoot, or do jumping jacks!

We realize this is a difficult time, but for those who can consider giving, the donation link is [here](#). 100% funds go directly to research. And, Lulu's Crew Team STXBP1 has met the requirements to receive matching funds from the University of Pennsylvania Orphan Disease Center. Our goal is to fund two seed grants, as we did last year; your contributions are important to this goal!

Last year, STXBP1 grants from the MDBR and Orphan Disease Center funded important research projects with Co-PIs Dr. Matthijs Verhage at Vrije Universiteit Amsterdam and Dr. L. Neukomm at University of Lausanne, as well as Dr. Zachary Grinspan at Weill Cornell for a 4-phenylbutyrate clinical trial pilot.

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>
- Jackie Steinberg (Alex's mom): <http://givingpages.upenn.edu/lulucrewJackie>
- Jennifer Clatterbuck (Emma's mom): <http://givingpages.upenn.edu/TeamSTXBP1>
- Charlene Son Rigby (Juno's mom): <http://givingpages.upenn.edu/lulucrewCharleneR>

To ride with Lulu's Crew Team STXBP1 you can register at: <https://www.milliondollarbikeride.org/registration>! Be sure to sign up for Lulu's Crew/Team STXBP1 and select "with fundraising." Once you have registered, forward your confirmation email to [info@stxpb1disorders.org](mailto:info@stxpb1disorders.org) to set up your fundraising page!

## HOPE Project for STXBP1

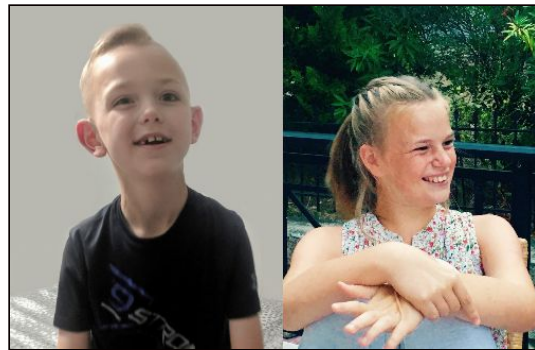


insurmountable challenges every day in caring for their STX'er, but who still, somehow, manage to keep everything running smoothly and ensure their children feel safe and loved. It is easy to become discouraged about day to day life and for what the future may hold, so at the end, we have a special message of hope for our community.

The HOPE Project is a labor of love and we look forward to sharing it with you. We will post the video link on our STXBP1 Disorders page on Facebook when it is ready! We hope you can feel every ounce of love that was poured into it as you watch.

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## Funds in Honor of Spencer James Manwaring & Elena Ramnialis



We are honored to announce memorial funds for two of our STXBP1 angels, Spencer James Manwaring 2010-2019 and Elena Ramnialis 2004-2020. Both funds were created in collaboration with the families, and will fund research to find a cure for those with an STXBP1 disorder.

We honor the memory of Spencer and Elli, and all of our STXBP1 angels everyday in everything we do.

Spencer's Fund: <https://donorbox.org/spencermanwaringfund>

Elena's Fund: <https://donorbox.org/elli-s-smile-fund>

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## Researcher Spotlight: Ganna Balagura



We plan to feature researcher updates in our quarterly newsletter. This issue, we feature

Session.

Ganna Balagura is working on a genetic therapy approach for STXBP1 using non-coding RNA molecules (SINEUP). At the University of Genoa, Ganna was able to identify several SINEUPs that increase expression of STXBP1 in a cell line. She presented this work at AES in Baltimore, and was awarded a blue ribbon for her research project. Ganna is now at Matthijs Verhage's lab for a couple months continuing work to validate these SINEUP candidates in human neurons.

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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. 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.

We are excited to offer a couple of fun, socially distanced options for fundraising within your local area, but we need your help. Email [melissa.hioco@stxbp1disorders.org](mailto:melissa.hioco@stxbp1disorders.org) if you are interested in launching a fun fundraiser in your area, in which the whole family can get involved. Students who need community service hours are encouraged to get involved, and we are happy to sign off on the hours.

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](mailto:melissa.hioco@stxbp1disorders.org) with your ideas!

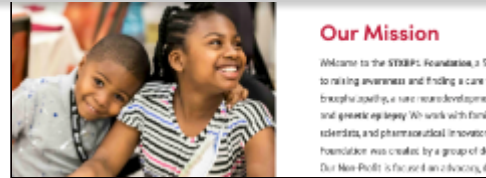
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## Join the STXBP1 Registry



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](#).

## Updated STXBP1 Foundation Website



If you haven't checked out the [new website](#) we launched in January, check it out! It is easier to navigate, and has expanded content including information on STXBP1 researchers and projects. We welcome your [feedback](#) on the new site.

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## Upcoming Research Conferences



The Global Genes [RARE Drug Discovery Conference](#) (6/11-12) and the [Epilepsy Foundation Pipeline Conference](#) (8/26-27) have both gone virtual. Charlene Son Rigby will represent STXBP1 at both of these meetings. If you are interested in attending, please visit their registration sites or [contact us](#).



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