Happy Summer from the STXBP1 Foundation!

Welcome to our July newsletter. Read about STXBP1 Summit+, our first FDA meeting, new Foundation Board Members, the STXBP1 World Map, fundraising driven by our amazing families and recent grants!

STXBP1 Summit+ 2022

Don't miss it!

Get ready for the STXBP1 Summit+ taking place August 19-20, 2022 in Philadelphia!

The STXBP1 Summit+ will bring researchers, clinicians, and families together to review and understand the latest work on STXBP1 disorders, provide networking opportunities for researchers, clinicians and families, connect researchers with families and the patient community, and provide social opportunities for families.

In addition, there will also be opportunities to sign up for research studies and contribute samples to biorepositories - important for advancing research and clinical trial readiness.

Registration is open through July 24! Learn more and register here**.**

Can't make it to Philly? We got you covered. All sessions will be livestreamed online and recorded to watch at your own pace afterwards.

STXBP1 World Map

STXBP1 disorders is a rare genetic disease that affects patients worldwide. We've built a map as a visual representation of where our community is located across the globe. **Patient families, please sign up for our Contact List** (at this link) **to be added to the map.** We have 349 families so far!

In addition to showing the variety of locations where STXBP1 patients live, this information helps us to connect you to research studies in your geographical area and Foundation activities. Listings on the map can be anonymous; you can choose to show age and/or first name, or only a location pin. **View the** STXBP1 World Map here**.**

$85K Raised at FC Builders Golf Tournament

Thank you to FCL Builders for selecting the STXBP1 Foundation for their annual charity golf tournament, and to Helen Szazy, a fellow STXBP1 parent, who helped organize the event! Through the generosity of the golfers and sponsors of the event, roughly $85,000 was raised which will fund a new project on biomarkers.

FDA Listening Session on STXBP1

The STXBP1 Foundation requested its first meeting with the U.S. Food and Drug Administration (FDA), and held a Listening Session with the FDA on April 12 2022. The

goal was to educate the FDA on STXBP1-related disorders including a broad overview of the lived experience of people with STXBP1-related Disorders, the requirements for daily caregiving of affected individuals, and treatment priorities.

The bottom line, which was presented, is that new treatments are urgently needed for STXBP1 patients who currently have very limited options.

Read the summary here.

Lulu's Crew at MDBR: 4th Year Raising Money for STXBP1 Research

The Million Dollar Bike Ride took place on Saturday, June 11. The final tally is still being calculated but Lulu's Crew Team STXBP1 again raised over $150,00! Props to our organizing family Ben and Erin Prosser, parents of Lucy, and all of our riders who have diligently been raising funds towards STXBP1 research for 4 years.

With these funds, two new grants will be available for STXBP1 research through University of Pennsylvania's Orphan Drug Center! More information and the request for applications will soon be posted on the ODC site. Make sure your favorite researcher is aware of this opportunity!

SCIENCE + LOVE = CURE

SCIENCE + LOVE = CURE shirts are back!!

Back by popular demand and just in time for the STXBP1 Summit+ in August!

SCIENCE + LOVE = CURE shirts that are not personalized can be ordered here.

AND...for our families, we again have the option to personalize a campaign and shirt for your STX'er! Go to this link and begin with the template provided.

New Board Members: Cristina Brennan & Jared Barnum

We are delighted to welcome Cristina Brennan and Jared Barnum as the newest STXBP1 Foundation board members! Cristina is now the STXBP1 Foundation Secretary. Jared holds a new position as the Director of Technology. We are tremendously excited to have these two amazing and passionate STXBP1 parents on the board! Learn more about Cristina, Jared and the rest of the foundation board members on the Team page on our website.

Science Saturdays with Jim

The Foundation is excited to present Science Saturdays with Jim, our Scientific Director. As we get closer to the Summit+ conference in August Jim will make a couple of short videos to provide some background information that he hopes will help you better understand some of the topics that will be discussed during the Summit.

Topics he will cover include **what are natural history studies and why they are important**, **an introduction to the field of gene therapy**, **what do scientists and drug developers mean when they talk about preclinical studies**, and **the types of clinical studies and how they are run.**

Join us on our FB page, Stxbp1 Disorders, every Saturday from now until the Summit+ for Science Saturdays with Jim!

Are You All In?
Building STXBP1 Natural History

Natural history studies are very important to rare disease communities like STXBP1. To improve care and develop effective treatments for STXBP1 disorders, we need a detailed understanding of patient symptoms over time. Natural history studies allow us to capture this important information.

STXBP1 affects our children in different ways, so it's important that we have broad representation of our patients.

From now until end of September -- STXBP1 Awareness Month -- the STXBP1 Foundation will be giving out $25 Amazon gift cards for those who enroll into the Contact List and at least one of these Natural History Studies: Simons Searchlight, Ciitizen, or RARE-X.

Send us a snapshot of verified enrollment to naturalhistory@stxbp1disorders.org. Gift cards will be sent in October.

Funding Award: Dr. Liz Heller

The STXBP1 Foundation is excited to announce a two-year **$154,753** grant to Elizabeth Heller, PhD at University of Pennsylvania Perelman School of Medicine. Dr. Heller's grant is titled "Editing the Epigenome: Curing STXBP1/MUNC18-1 Heterozygosity." Dr. Heller studies regulation of genes, and her work is important for STXBP1 to understand how STXBP1 gene expression might be regulated. Dr. Heller, excited you are joining the STXBP1 research community!

Learn more about the STXBP1 Foundation's recent grant funding here.

New Neurogenetics Clinic at Children's Hospital Colorado

Children's Hospital Colorado

We are thrilled to have launched a Neurogenetics Multidisciplinary Clinic at the Children's Hospital Colorado. We are partnered with 3 other rare disease organizations to launch this new clinic to provide comprehensive patient care while better informing research on each disorder.

The clinic is being held once a month. Learn about the Colorado clinic here including how to schedule an appointment.

Funding Award: Dr. Zach Grinspan & Ravicti Trial

The STXBP1 Foundation has provided a one-year **$50,000** grant to Zachary Grinspan, MD MS at Weill Cornell Medicine. Dr. Grinspan's grant is titled "Phenylbutyrate for STXBP1 Encephalopathy – 1 year follow-up evaluation." Dr. Grinspan is leading the ravicti / 4-phenylbutyrate repurposed drug clinical trial.

This grant will support 1 year follow up for all patients in the initial clinical trial who continue to receive the medication. Patients will participate in quarterly visits which will help in understanding longer term impacts of 4-phenylbutyrate and provide data on exploratory clinical outcomes which will help in our broader clinical trial readiness preparation. Dr. Grinspan, thank you for your important work to advance therapies for STXBP1!

Learn more about the STXBP1 Foundation's recent grant funding here.

6th Annual Move to Cure STXBP1 Disorders

The 6th Annual Move To Cure STXBP1 Disorders event will take place September 24 - 25. 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 get to 9! 9 miles because STXBP1 is on the 9th chromosome!

The purpose of this challenge is to raise awareness of STXBP1 Disorders, fundraise for STXBP1 research, and bring the community “together” for an important cause and some fun! Keep it simple, and get Moving for a Cure!

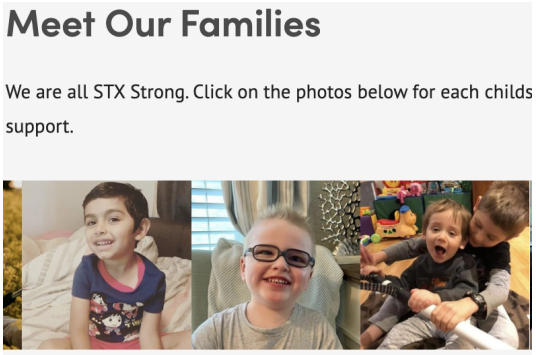
Last year, we raised over $80,000 for research, and 250+ people around the world participated. We can't wait to see how the community shows up this year! **#move2cureSTXBP1**

Sign up here: Move to Cure Virtual Challenge sign up

Buy custom shirts from Bonfire here: Team shirt

Questions? Contact Allison Michels.

STX Strong Sundays



The STXBP1 Foundation is excited to announce a new, weekly series spotlighting an STX'er and their family every Sunday, called STX Strong. A total of 28 STXer's have been featured to date. Please follow along on Facebook and Instagram here: #stxstrong

We recognize how meaningful the individual introductions are for both our families and their personal support communities. Donors have also expressed appreciation for the opportunity to learn a little bit about the individuals whose lives their generosity impacts. Make sure to follow us on social media, so you don't miss the chance to meet any of these special STX'ers and their families.

You can "Meet our Familes", and learn how you can participate here.

Together we are STX Strong!

STXBP1 Summit+ Family Travel Scholarships Awarded

The STXBP1 Foundation has awarded travel assistance scholarships to 4 STXBP1 families, who met the applicable criteria for the scholarship funding. There were an overwhelming number of applications, and we wish we could fund more. We are happy to be able to provide this opportunity to these 4 STXBP1 families. We look forward to meeting you in Philly!

Your Feedback Please!

STXBP1 Community, the STXBP1 Foundation wants your input on needs, priorities, feedback and how we can better serve the community.

Please answer this short survey.

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)
- Michigan
- Texas
- California

Call for Volunteers!

Share your talents and time while helping to further the mission of the STXBP1 Foundation. We are looking to fill the following volunteer positions.

- **Volunteer Lead** - Recruit and manage volunteer network and to assist the Foundation with areas of need. Hours will vary, anticipating commitment of 10 hours per month
- **Community Communication Volunteer** - Prepare monthly newsletter. Approximately 1-2 hours per week
- **Social Media Volunteer** - Promote content on Social media platforms. Approximately 10 hours per month
- **Fundraising Committee Volunteers** - Involved with developing fundraising ideas, implementing fundraising initiatives including grants, and collaborating to promote and implement fundraising events. Approximately 10 hours per month

To volunteer or to get more information on any of the above volunteer opportunities, please contact Melissa Hioco.

Calling all Grandparents to the Grands Society!

Grandparents, join the Grands Society!

STXBP1 Grandmother, Helen Rigby, has started 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.

STXBP1 Now Accepts Donations of Stocks and Cryptocurrency

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 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 within the U.S.A.

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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