2023 Impact Report

STXBP1disorders

From Our President

Dear STXBP1 Community,

Charlene Son Rigby
President & Mom

When we embarked on the **Fast Forward** strategic plan in 2023, this STXBP1 community jumped in, grabbed the wheel, and stepped on the gas.

As we take a moment to check our rear view mirror, we are filled with gratitude for how far we have come in the last year alone.

Together, we have achieved significant milestones that accelerated our progress, and I wish to thank you—our families, funders, and partners, who continue to play an important role in this journey toward our goals. This past year we invested more than $1.5 million in grants to propel precision therapies for STXBP1 and launch our STARR Natural History Study as our community prepares for clinical trials ahead.

We simply could not do all of this without you. You created personal fundraisers, participated in our annual signature fundraising effort, Move to Cure, partnered with Lulu's Crew for the Million Dollar Bike Ride, sponsored and attended our STXBP1 Summit, invested your personal contributions through unique initiatives like FLOURISH, and used your voice and personal platforms to encourage others to join our journey.

The road ahead is paved with the promise of progress toward treatments and a possible cure for STXBP1-related disorders. We are more driven than ever to improve the lives of our loved ones, and we are so grateful to be fueled by your generosity.

With gratitude,

Charlene Son Rigby

2023 Impact Report
Top 10 Achievements

STXBP1foundation

1 Fast Forward

Launched new STXBP1 **Fast Forward 3-Year Strategic Plan**.

2 Disease Concept Model

Published the first peer-reviewed STXBP1 Disease Concept Model documenting our patients' **most significant symptoms** and their impact.

3 European Research Roundtable

Held first Research Roundtable in Europe bringing together **over 100 researchers** and clinicians in Milan.

4 STARR Natural History Study

Launched first longitudinal Natural History Study for STXBP1-related disorders **enrolling at least 100 patients at 4 sites** across the USA.

5 FDA Listening Session

Orchestrated first externally-led Patient-Focused Drug Development meeting with the FDA for STXBP1, as **288 attendees** joined in to discuss patient experiences and the **urgent need for treatments.**

FDA

6 Global Connect

Exceeded 20 countries in the STXBP1 Global Connect, our coalition of STXBP1 foundations and country liaisons.

STXBP1 Global Connect

20+ Countries

7 Patient Census

Initiated an STXBP1 patient census, which has counted **over 973 patients in 48 countries** around the world.

973 patients

48 countries

8 Grants & Research Funding

Awarded over **$1.5M** to date for Patient Studies and Translational & Pre-Clinical Research projects around the world.

9 Patient Assistance Grants

Awarded over **$20K** to date for support of STXBP1 patients and their families.

$20K

10 STXBP1 Summit + Annual Conference

Held our first Family Meeting in Colorado with over 300 attendees.

STXBP1 Summit+

Our Community

Throughout 2023, our community came together in remarkable ways, showcasing the power of unity and shared purpose.

Together we advocated for our families and loved ones, amplified our voices to raise awareness, and, most importantly, forged connections with each other electronically and in person.

Advocacy

Awareness

Connection

Advocacy

Meeting with FDA

The STXBP1 Foundation held a **Patient-Focused Drug Development (PFDD) meeting**, our first public meeting with the FDA, in October. This was a very special opportunity to inform the FDA of the lived experience with STXBP1 for patients and our families, and elevate our voices to emphasize the urgent need for truly impactful therapies. **More than 288 people attended**. We are grateful to Biogen, BioMarin, Encoded Therapeutics, and Horizon (now Amgen), for their generous partner support, and we are especially grateful to the families who so openly shared their compelling personal stories about the challenges of STXBP1 disorders.

American Epilepsy Society

STXBP1 was well represented at the American Epilepsy Society annual meeting, where experts in neurodevelopment disorders and epilepsies unite to network, collaborate, and share research. This year **STXBP1 was represented in 13 scientific posters**— a leap from five in 2019!

13 Scientific posters on STXBP1

STARR Natural History Study

The STARR Natural History Study for STXBP1 held its first-ever meeting with all clinical sites, to discuss the first set of patients seen in the study in 2023. **The four sites were launched in 2023**: Children's Hospital of Philadelphia, Children's Hospital Colorado,Baylor Texas Children's Hospital, and Weill-Cornell in New York.

Awareness

Awareness Ambassador

STXer Lily Kahrl, 17, and her family have made a big splash in their community to spread awareness about STXBP1. A member of the Wellesley High School swim team, Lily shares a special bond with her teammates and peers, who have rallied around her and her family's efforts to elevate awareness for STXBP1 Disorders.

Rare in Times Square

Our community reached new heights on Rare Disease Day 2023 as beautiful STXer Emma represented STXBP1 awareness in Times Square in partnership with Beyond the Diagnosis.

> **Her face represents her community in medical universities, government facilities, and research labs all over the United States, educating all who see it. My daughter is finally completely visible, her disease has a name, and she is home.**
>
> **Emma's mom**

Going to bat for STXBP1

During STXBP1 Awareness Month in September—aptly the ninth month of the year for the ninth chromosome, which STXBP1 affects—Major League Baseball player Shelby Miller, who played for the Dodgers in 2023, was featured in the news, where he raised awareness about STXBP1 Disorders, sharing his family's story about their 3-year-old STXer Kyler.

Connection

2023 STXBP1 Summit

The STXBP1 community is a source of strength and support for families navigating this journey, and when we come together under the same roof, we are united by a common purpose to support our loved onesand each other. **The 5th Annual 2023 Summit in Colorado** was an unforgettable gathering that brought together **more than 300 attendees** in person and countless more online, comprising families, esteemed researchers, clinicians, and industry partners. Together we are stronger and faster. Abundant thanks to our generous sponsors!

Through the generosity of philanthropic donations, STXBP1 Foundation had the honor of awarding **10 family scholarships** to parents and caregivers who may have otherwise faced barriers to attending the Summit.

> **This is a family we never knew we would need, but we're so glad to have.**
>
> **Heather Jones**

Monthly Newsletter and Podcast

In 2023 we enhanced regular communications with our monthly Newsletter and newly-launched podcast: We launched a podcast! Now, anyone anywhere can catch a 10-minute monthly update for the STXBP1 Community through our STXBP1 Science + Love = Cure Podcast. Available on **Apple podcasts** or by video on **YouTube**, the podcast offers an overview of the most important relevant topics for our community each month.

Counting on Each Other

In 2023, **STXBP1 Global Connect**, an international network of parents and patient advocacy groups from more than 20 countries, collaborated with STXBP1 Foundation around a common goal—launching a census! Knowing where patients and families are worldwide helps our community plan for clinical care and future studies, especially as we advance toward possible clinical trials. As of December 31, we counted more than 973 patients in 47 countries around the world—which is incredible to think we had only a couple hundred of known cases when the Foundation was established in 2017.

STXBP1 Global Connect

Research

Our determined STXBP1 Community helped us buckle up and accelerate progress in 2023.

Together, through the generosity of this community of partners, donors, and families, we accelerated our work to propel precision therapies and prepare for clinical trials.

The STXBP1 Foundation has invested $1.5M into research and around STXBP1-related disorders to date, with $760,655 in 2023 alone. As part of the STXBP1 Fast Forward 3 Year Plan, we have focused many of our efforts in 2023 toward preparing for clinical trial readiness, including supporting several clinic sites for the STXBP1 STARR Natural History Study.

$1.5M

Research Funding since 2017

$760,655

Research Funded by STXBP1 Foundation in 2023

STARR Shining Brightly

As the signal turned green for the **STARR Natural History Study**, our families didn't hesitate to step on the gas. In 2023, the STXBP1 community rose to the occasion in remarkable ways. Recognizing the importance of gathering patient data to help inform upcoming clinical trials, we initiated a multi-site longitudinal prospective Natural History Study to provide clinicians and researchers with comprehensive insights into our STXers.

Setting a goal to enroll 65 patients by year-end, we were thrilled to surpass expectations with nearly 100 patients enrolled by December 31. This achievement wouldn't have been possible without the generous support of philanthropic individuals who backed the FLOURISH initiative in 2022, and the ENDD Center at Penn Medicine and Children's Hospital of Philadelphia, enabling us to launch this study in its inaugural year.

ENDD
Center for Epilepsy and NeuroDevelopmental Disorders

The Children's Hospital of Philadelphia®

In 2023 we also celebrated

The first peer-reviewed **STXBP1 Disease Concept Model** was published, documenting our patients' most significant symptoms, and the impact the disorder symptoms have on them and their families.

The first ever **STXBP1 Research Roundtable** in Europe, was held in Milan. The meeting drew more than 100 researchers from around the world who shared what they're discovering. Collaborative efforts showcased at the meeting underscored the dedication to unraveling STXBP1 complexities and advancing treatment strategies. With ongoing natural history studies and growing international collaborations, the future holds promise for better understanding and managing STXBP1-related disorders.

Funded Research

Patient Studies

$20,000	**Elena Gardella, MD, PhD** Danish Epilepsy Centre for a study on Early Mortality in STXBP1 Encephalopathy
$165,001	**Zachary Grinspan, MS, MD** Weill Cornell Medicine/ New York-Presbyterian for STXBP1 STARR Natural History Study
$155,264	**Andrea Miele, PhD** Children's Hospital Colorado for STXBP1 STARR Natural History Study
$200,000	**Hsiao-Tuan Chao, MD, PhD** Baylor College of Medicine/ Texas Children's Hospital for STXBP1 STARR Natural History Study
$200,000	**European STXBP1 Consortium (ESCO)** Research consortium for European Natural History Study and Registry

Translation and Pre-Clinical Research

$20,000	**Alex Felix, PhD** University of Pennsylvania for Young Investigators Draft class for rare disease research co-funded with Uplifting Athlete

Riding for Research with Lulu's Crew/Team STXBP1

STXBP1 is grateful to Erin and Ben Prosser who in 2023 led Lulu's Crew/Team STXBP1 to surpass $745,000 total raised for STXBP1 research over five years.

In 2023, the Orphan Disease Center awarded $150,920 for two STXBP1 Research grants, split evenly to Wendy Gold, PhD at University of Sydney, and Xuebing Wu, PhD at Columbia University.

Clara Inspired's Generous Investment in a Little

In collaboration with Jackson Labs, we worked to create a specialized nonsense mouse model for STXBP1. The model mimics the effects of nonsense mutations, providing invaluable insights into the condition and potential treatment avenues. STXBP1 Foundation is grateful to Clara Inspired for their significant investment in this initiative, and for all the ways they accelerate progress.

Your Philanthropic Investments Hard at Work

STXBP1 Foundation is proud to fund grants to help enable investigator-led research to broaden our understanding of STXBP1 biology and develop safe and transformative therapies for STXBP1-related disorders. Through a rolling grant program, open to research investigators, we fund multiple types of projects aimed at addressing the diverse needs of the STXBP1 field. And we simply could not do the support of this community. Thank you!

Fundraising

Our deepest gratitude to those who generously donate to support the mission of the STXBP1 Foundation. We are immensely grateful to our donors, sponsors, and philanthropic partners.

Highlighting the hard work and determination of just a few of the many who held fundraisers in their communities.

A Cure for Charlie

Clara Inspired

FLOURISH

Giving Tuesday - Be the Love Campaign

Lucy's Light

Lulu's Crew/Team STXBP1
For the Orphan Disease Center at University of Pennsylvania School of Medicine Million Dollar Bike Ride

Miles for Maya

Move to Cure

Party with a Purpose

Teddy's Rough Riders/Teddy Strong

Voyage Toward a Cure

Partners

Baylor College of Medicine · Texas Children's Hospital · Children's Hospital Colorado Foundation · Weill Cornell Medicine · The Children's Hospital of Philadelphia · Penn University of Pennsylvania · ENDD Center for Epilepsy and NeuroDevelopmental Disorders

ciitizen · RARE-X · CURE EPILEPSY · COMBINED · Amsterdam UMC University Medical Centers · Università di Genova

The Arc Colorado · REN Rare Epilepsy Network · SIMONS SEARCHLIGHT · FILADELFIA · STXBP1 Global Connect · ESCO

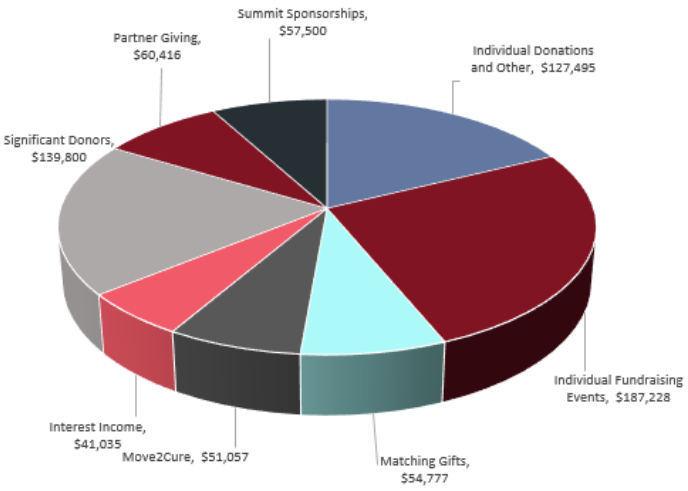
Financials & Transparency

Across the STXBP1 community, an extraordinary spirit of philanthropy thrives. Parents, caregivers, researchers, clinicians, and even strangers touched by our cause, demonstrate unwavering support through their generosity and advocacy. Their collective efforts, whether through fundraising events, donations, or spreading awareness, fuel our mission's progress.

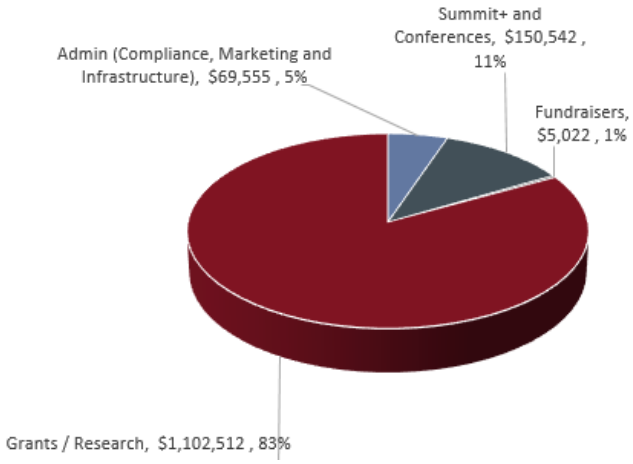
As a foundation, we take immense pride in our fiscally responsible practices, with over 94% of every dollar directly funding crucial research and family engagement and support. This remarkable dedication empowers us to drive forward with confidence, knowing that together, we are making a tangible difference in the lives of those affected by STXBP1-related disorders.

We are pleased to share the following overview of contributed revenue and expenditures for the calendar year 2023.

Donation by Category 2023



Expenditures by Category 2023



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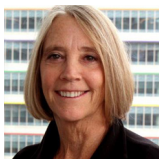

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STXBP1 Global Connect & Regional Groups

Learn about **STXBP1 Global Connect**, and in the US, join a **regional group** to connect with our network of regional support.

Volunteer

Have skills or interests you want to share?
STXBP1disorders.org/volunteer

Read Our Blog

Check out our **latest blog posts**!