STXBP1 disorders

Science + Love = Cure

Welcome to the March-April issue of the STXBP1 Newsletter

It's official - on June 10th Lulu's Crew Team STXBP1 rides again!

Join Us! (in-person in Philly or virtual on your Peloton/stationary/road/mountain)🚴‍♀️💜

This fundraising effort has raised more than $600K over the last four years to fund important research to advance STXBP1 research and therapy development.

Let's keep going strong! Registration is now OPEN HERE!!

STXBP1 Families join us this July in Colorado!

Get ready for the STXBP1 Summit+ Family Meeting July 22-23 at the Westin Westminster in Westminster, CO (just outside of Denver).

Early bird registration is available through 5/31. Registration closes on 6/22.
REGISTER HERE!

The STXBP1 Summit+ Family Meeting will bring researchers, clinicians, and families together to review and understand the latest work on STXBP1 disorders. Families will also have networking and social opportunities.

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

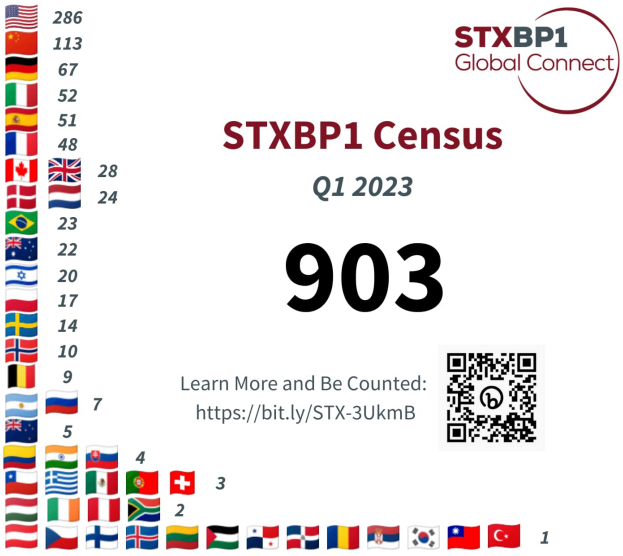
The first global census for STXBP1 is published!

903 patients from 45 countries!

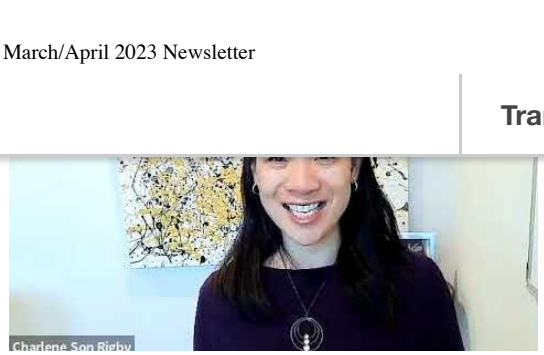
We know there are more STX'ers out there…and we will continue to work to find them!

STXBP1 parents join the Contact List HERE to be counted! An accurate count of STX'ers and where they live helps us support our patients and families, and plan for clinical care and studies.

Check out the news article HERE for more information on the census.



STXBP1 Podcast!



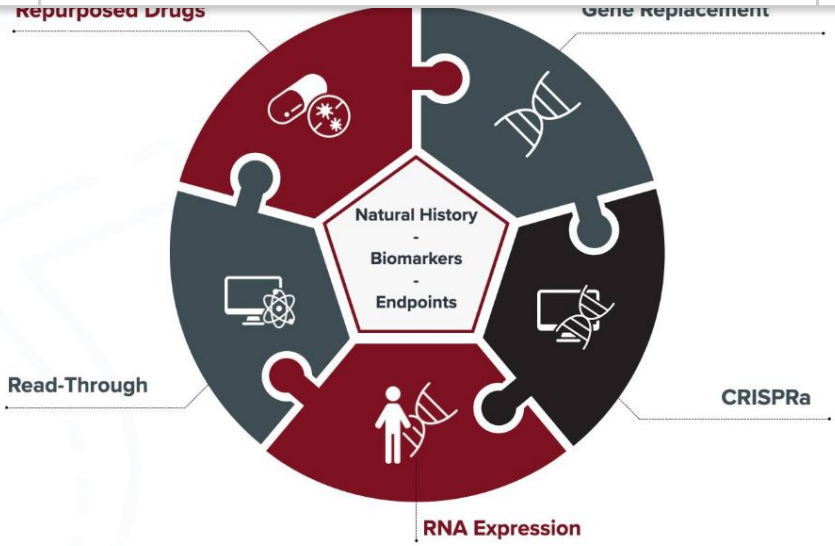
Science + L❤️ve = Cure 10 Minute Updates from the STXBP1 Foundation for the STXBP1 Community! Please tune in to the Foundation's new podcasts featuring Foundation President Charlene Son Rigby for updates and information on all things STXBP1. The 10 minute podcasts can be found on our YouTube channel HERE.

$25 Million Gift to Penn Medicine and CHoP Establishes Center for Epilepsy and Neurodevelopmental Disorders

STXBP1 dad, Ben Prosser, PhD, is the director of a new center at the University of Pennsylvania, which will further multidisciplinary research for STXBP1. The center was established with a $25 million funding by an anonymous donor!

The donation was made to Penn Medicine and Children's Hospital of Philadelphia (CHOP) and will establish the Center for Epilepsy and Neurodevelopmental Disorders (ENDD), to accelerate collaborative research in genetic therapies for neurodevelopmental disorders.

Dr. Prosser is dad to STX'er Lucy, and is an associate professor of Physiology at the University of Pennsylvania's Perelman School of Medicine. When Lucy was born in 2018, Dr. Prosser adjusted the focus of his research career to study treatments for STXBP1. Read more about the ENDD Center here.
(Pictured above is Ben and Erin Prosser with daughter, Lucy)



STXBP1 Fast Forward Launches

Now is the Time for the STXBP1 Community to Activate...Now is the Time to Move STXBP1 Fast Forward! Now, the STXBP1 Foundation is launching a new three-year plan: **STXBP1 Fast Forward.**

Research is progressing quickly and we are optimistic that viable therapeutics are possible because of these four important learnings:

📌The root cause is known - changes in the STXBP1 gene cause STXBP1 disorder

📌We have a strong understanding of STXBP1's biological function in the brain

📌We have a much better understanding of the clinical picture of STXBP1 disorders now than we did even 3 years ago.

📌Restoring levels of the wildtype STXBP1 protein reverses some disease-relevant symptoms in mice

With 9 therapies (and counting) in the pipeline, we need to move STXBP1 Fast Forward toward clinical trials and approved treatments...and a cure! Join us...as together we move STXBP1 Fast Forward!

Learn more about the Fast Forward 3 year plan HERE

Rare in Times Square

Look who's at the center of the world. Say hi to **Emma**. Emma is living with an STXBP1-related disorder. Emma's portrait, by Lawrence Gardinier, is one of dozens featured in **Times Square** in February to celebrate **Rare Disease Day on February 28th**.

See Emma's portrait and visit Rare in Times Square to learn more.

Rare Disease Day is an international day recognizing the rare disease patient and their support networks. The NIH Rare Disease Day was started as a way for the public, patient advocacy groups, and the scientific community to bring awareness of rare disease and the challenges they present.

See HERE for information from the event. Pictured is STXBP1 Foundation Vice President Jackie Steinberg with Rare Disease advocates and research partners from RARE-X and Ciitizen.

In the US, the month of April is National Volunteers Month. The STXBP1 Foundation would like to take this moment to thank all the volunteers around the world that dedicate their time and energy into spreading awareness, raising funds and contributing to efforts in improving the lives of those in the STXBP1 community.

Click here to sign up to volunteer to help the STXBP1 Foundation.

Important Notice for Families Receiving Medicaid!

Post-Covid Medicaid eligibility redeterminations are re-starting and every person (both children and adults) on Medicaid will have their eligibility redetermined.

- Avoid gaps in coverage and find your states timelines here: https://www.medicaid.gov/.../ant-2023-time-init-unwin...
- To learn more, read the CMS Toolkit here https://www.medicaid.gov/.../unwinding-comms-toolkit.pdf
- And the Epilepsy Foundation page here: https://www.epilepsy.com/.../state-medicaid-eligibility...

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The STXBP1 Brand Gets a Fresh Update

Read the update rationale and design details in our recent blog post. You can even download the Style Guide and use our updated logos right away from the new Brand page.

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