



STXBP1 Disorders

Leading the Charge For a Cure

Welcome to the January issue of the STXBP1 Newsletter.

2022 YEAR IN REVIEW

With your support, we are advancing research to accelerate therapies. In addition, we are building community and supporting our families on this challenging journey



- STXBP1 CLINICAL TRIAL FOR 4-PHENYLBUTYRATE, WITH INITIAL RESULTS REPORTED AT AMERICAN EPILEPSY SOCIETY MEETING
- DISEASE CONCEPT MODEL FOR STXBP1 PUBLISHED
- FDA LISTENING SESSION TO EDUCATE THE FDA ON STXBP1 & OUR COMMUNITY'S LIVED EXPERIENCES



NEW STXBP1 FOUNDATION BOARD OF DIRECTOR MEMBERS, JARED BARNUM & CRISTINA BRENNAN



FLOURISH ART AUCTION & FUNDRAISING CAMPAIGN

\$1.2M RAISED
TO FUND CLINICAL TRIAL READINESS & NATURAL HISTORY STUDIES



300+
PARTICIPANTS FROM AROUND THE WORLD MOVING IN THE ANNUAL MOVE TO CURE STXBP1 EVENT

DEBUT OF STXBP1 GLOBAL CONNECT WITH 15 COUNTRIES REPRESENTED



STXBP1 SUMMIT+ RESEARCHER & FAMILY MEETING IN PHILADELPHIA



LAUNCHED SCIENCE SATURDAYS SERIES WITH JIM, OUR SCIENTIFIC DIRECTOR

27 PATIENT STEM CELL LINES

- 15 IN SIMONS SEARCHLIGHT
- 2 IN THE NEW STXBP1 FOUNDATION BIOREPOSITORY



Driven by science. United by hope.



125 PATIENTS ENROLLED IN CITIZEN NATURAL HISTORY STUDY & LAUNCHED RARE-X NATURAL HISTORY STUDY

2022 was an amazing year for the STXBP1 Foundation and Community! Please view our [2022 Impact Report](#).

Hold the Date! 2023 STXBP1 Summit+ Family Meeting in Colorado



Join us this July 22-23, for our 4th Annual STXBP1 Summit+ Family Meeting at the Westin Westminster in Westminister, CO (just outside of Denver).

STXBP1 families make your hotel reservations [here](#).

This year's in-person event is in partnership with the Children's Hospital Colorado (CHCO) in Aurora, CO. Families will have the opportunity to connect with other STXBP1 families, learn the latest information on STXBP1 including research updates, participate in break out sessions for families, and have fun during social gatherings.

STXBP1 Clinic Day - a limited number of appointments are available on Friday, July 21, at the Neurogenetics Multidisciplinary Clinic at CHCO. Please contact Nurse Coordinator Tristen Dinkel at 1(707)777-7453 or BNDP@childrenscolorado.org, to schedule an appointment.

Summit registration information coming soon.

2023 Children's Hospital of Colorado Clinic Funded



Due to the great success and continued positive feedback from families who have attended the Neurogenetics Multidisciplinary Clinic at the Children's Hospital Colorado, we are thrilled to report that we have continued to fund this important resource for our families through 2023!

The STXBP1 Foundation has partnered with 3 other rare disease organizations (SLC6A1Connect, Ring14 and Project 8p) to continue supporting this all-encompassing clinic to provide comprehensive patient care while better informing research on each disorder. Follow the link above for our 2023 Summit+ to learn about making an appointment for your STXBP1 patient at the clinic.

Learn about the Colorado clinic [here](#)



STXBP1 Foundation is participating in the Xcelerate RARE: A Rare Disease Open Science Data Challenge.

Xcelerate RARE is bringing together researchers and data scientists in a collaborative and competitive environment to make the best use of STXBP1 patient-provided data through the RARE-X - STXBP1 Disorders Data Collection Program.

RARE-X anticipates that some of the outcomes of Xcelerate RARE might be:

- Shortening the diagnostic odyssey with predicted diagnosis
- Identifying previously unrecognized symptoms associated with some of these rare diseases
- Generating useful insights for advancing therapeutic research

Enroll in the STXBP1 Disorders Data Collection Program today and not only **Be Counted**, but **Be Seen**!

Deadline Alert! Enroll in the STXBP1 Disorders Data Collection Program today and complete the surveys before March 30 so the data you provide will be included in the the Challenge.

To enroll, go to: stxbp1.rare-x.org/

To learn more about the Rare Disease Open Science Data Challenge, go to: rare-x.org/open-science-data-challenge/

Have questions on how to register? Contact naturalhistory@stxbp1disorders.org for help!

