WAYS TO SUPPORT

- Meet the STXBP1 kids and their families by joining our Facebook communities and on stxbp1disorders.org
- Participate in natural history studies to accelerate understanding of STXBP1 and development of better treatment options. Register for the Simons natural history study: simonssearchlight.org.
- Join our family contact list for parents and primary caregivers: stxbp1disorders.org/contact-list
- Subscribe to the STXBP1 newsletter: stxbp1disorders.org/newsletter
- Read our blog and contribute your thoughts
- Share this brochure with your friends and family
- Follow us on Social Media to learn about the latest news and research: Facebook, Twitter & Instagram
- Volunteer your time and talents to the STXBP1 Foundation: stxbp1disorders.org/volunteer
- Donate to STXBP1 research: stxbp1disorders.org/ways-to-give
- Join our community, support one another and encourage other families as this is what keeps us going

“Join our community, support one another and encourage other families as this is what keeps us going”
WHAT IS STXBP1 DISORDER?

STXBP1 disorder is an autosomal dominant disease, resulting from de novo (new or spontaneous) mutations in the STXBP1 gene, which affects the brain and nervous system, due to impairment of transmission between nerve cells. Patients with the disorder typically have some of these symptoms: early onset epilepsy, global delay, cognitive impairment (mild to profound), movement disorders, and autism spectrum.

Signs & Symptoms

Seizures typically begin at around six weeks but may also begin anywhere from 1 day to 13 years. Symptoms may include: early-onset epileptic encephalopathy, global developmental delay, feeding difficulties, gross motor, fine motor and other movement difficulties. Intellectual disability and autism features are also common. Some patients receive other diagnoses such as cerebral palsy. While most patients are nonverbal, some families report their children learning to speak and/or sign.

Emma’s Story

Emma was born August 26th, 2008 after an uneventful pregnancy and a perfect delivery. I had no reason to expect anything was wrong and had dreamed of the moment I would look into my little girl’s eyes and feel a connection unique to the one I shared with my two sons. She had Apgar scores of 9 and 9. She didn’t get a perfect score only because she didn’t cry. Instead, she looked at me and around the room in wonder at this new world she was seeing. She latched on and nursed until she fell asleep. It was a cherished, albeit brief moment of bliss. As her brothers came in to meet her, she jerked hard in my arms and started crying. It took a good 15 minutes to calm her and a knot began to form in my stomach. So began a life I never expected and a 5 ½-year-long fight to find out what was making my little girl sick. Her symptoms included severe epilepsy, infantile spasms, GERD, kidney reflux, a malrotated bowel, failure to thrive, and a significant loss in skills with illnesses. She stumped doctors in 4 different states, including NIH’s Undiagnosed Disease Program, until a research study being performed by a lab in Arizona discovered the gene responsible: STXBP1.

Emma’s specific variant has not been reported to date so there is no other child I can compare her to. Each year holds at least one surprise crisis and hospitalization, sometimes more. We have accepted we don’t have all the answers, and have chosen to do what we can to help find a cure and be thankful for and enjoy each day we have with Emma.

Who are we?

The STXBP1 Foundation is comprised of a group of parents dedicated to raising awareness of STXBP1-related disorders among parents, physicians, scientists, and pharmaceutical innovators. We hope that our work will lead us to better understand the progression of STXBP1 and one day lead to a cure.

Diagnosis

STXBP1 diagnosis is made through molecular genetic testing, through a panel test, exome testing or rarely chromosomal microarray analysis.

Incidence

The disorder occurs in countries, populations, and ethnic groups around the world. The total number of STXBP1 patients diagnosed to date based on genetic testing is estimated at 1000 people worldwide. The estimated incidence of STXBP1 is 1 in 30,000, although the true prevalence of the disease is unknown, as many cases go under- or misdiagnosed. Males and females have equal risk for the disorder.

References

