WAYS TO SUPPORT

• Meet the STXB1 kids and their families by joining our Facebook communities and on stxbp1disorders.org by reading “Our Stories”
• Register your family at simonssearchlight.org to join our registry
• Read our blog and contribute your thoughts
• Share this brochure with your friends and family
• Follow us on Social Media: Facebook, Twitter, Pinterest, & Instagram. There you will learn about the latest news and research
• Donate to STXB1 research
• Understand that your support is crucial and appreciated
• 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”
Who Are We?

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

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 Apгар 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 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.

Diagnosis

STXBP1 diagnosis is made through molecular genetic testing, through a panel test, exome testing or chromosomal microarray analysis. The genetic testing results would identify a pathogenic heterozygous variant in STXBP1, or a contiguous gene deletion that includes STXBP1 and possibly adjacent genes.

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 300–400 people worldwide. The estimated incidence of STXBP1 is 1 in 90,000 based on a 2016 Danish study, although the true prevalence of the disease is unknown, as many cases go under- or misdiagnosed (Stamberger et.al. 2016).

References