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.

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


Treatment

There are currently no curative or disease specific treatments for STXBP1 disorders and management of the disorder is based on symptoms or supportive measures. Antiepileptic drugs (AEDs) are used to treat seizures but there is no single anti-seizure medication found to be effective for the disorder and management is individualized. For some, multiple AEDs are needed for adequate seizure control but approximately 25% of patients will not gain seizure control with AEDs. Specialists include, but are not limited to, neurologists, physiatrists, dieticians, gastroenterologists, ophthalmologists, physical and occupational therapists and speech pathologists.

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.

“Join us as we endeavor to better understand the progression of STXBP1. We hope this work will one day lead to a cure.