WAYS TO SUPPORT

Register

Understanding the natural history of *STXBP1* is essential to discovering improved treatments. Whether it is a repurposed drug or a novel therapy, validation in a controlled manner is paramount to understanding efficacy and consequently improving the lives of our *STXBP1* patients.

Families can register with Simons VIP by visiting simonsvipconnect.org. Caregivers need only supply a genetic report confirming diagnosis to get started.

Participate in Research

Participating in or initiating research is essential to finding better therapies. Research on *STXBP1* is grossly underfunded; your support is critical to changing the current paradigm.

Spread Awareness

STXBP1 is rare and our families often go for years without the correct diagnosis. Your support in understanding the need for genetic analysis and communicating with your colleagues and patients is important to getting our families the information they so desperately need.

Contribute

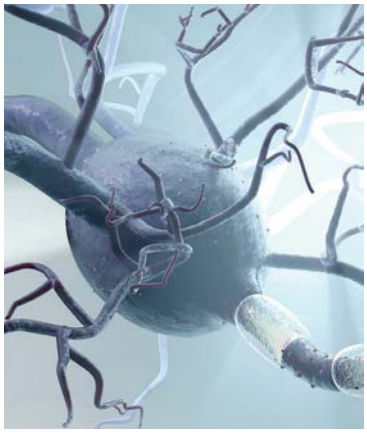
Whether it is your time or your money, this rare disorder represents an opportunity to make a meaningful and profound impact on *STXBP1* patients' lives. We hope you will support us today!

STXBP1 Disorders

www.stxdisorders.org
stxbp1@gmail.com

STXBP1 Disorders

Lead the Charge For a Cure



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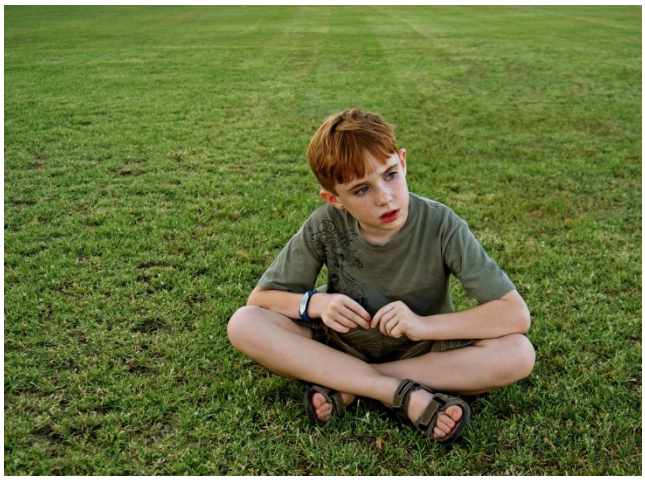
WHAT IS STXBP1 DISORDER?

STXBP1 disorder is an autosomal dominant disease, resulting from *de novo* 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.

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).



Signs & Symptoms

The median age of onset of seizures is six weeks (range 1 day to 13 years). Seizure types can include infantile spasms; generalized tonic-clonic, clonic or tonic seizures; and myoclonic, focal, atonic, and absence seizures.

Epilepsy syndromes can include: Ohtahara syndrome, West syndrome, Lennox-Gaustaut syndrome, and Dravet syndrome. Five percent of *STXBP1* patients do not exhibit seizures.

The clinical spectrum of *STXBP1* is heterogeneous and broad, with features overlapping other genetic disorders including: *SCN1A*, *MECP2*, and *KCNQ2*. 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 a diagnosis of Cerebral Palsy as the cause of disease. 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 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.

Treatment

Commonly used antiepileptic drugs (AEDs) are phenobarbital, valproic acid, and vigabatrin. In an estimated 20% of individuals, two or more AEDs are used in combination. Approximately 25% of patients do not respond to AED therapy. Severe dystonia, dyskinesia, and choreoathetosis can be treated with monoamine depleters or dopaminergic agents (Khaikin et. al.2016).

Supportive Care

Because patients with *STXBP1* disorder have a wide range of clinical manifestations and functional challenges, they are best followed by a multidisciplinary team. Many patients benefit from physical, occupational, feeding and speech therapies.

Long-Term Prognosis

Due to the rarity of and newness of molecular genetic testing for *STXBP1*, at this time only anecdotal information exists on long-term survival. Some *STXBP1* patients are in their 20's, 30's and even 50's.

References

Stamberger H., et. al. STXBP1 encephalopathy: A neurodevelopmental disorder including epilepsy. Neurology. 2016. 86(10): 954-62.

Khaikin Y., Mercimek-Mahmutoglu S. STXBP1 Encephalopathy with Epilepsy. GeneReviews Seattle (WA). 1993-2017.