WAYS TO SUPPORT

- Meet the *STXBP1* kids and their families by joining our Facebook communities and on www.stxdisorders.org by reading "Our Stories"
- Register your family at www.simonsvipconnect.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 *STXBP1* research
- Understand that your support is crucial and appreciated
- Join our community and support one another and encourage other families as this is what keeps us going

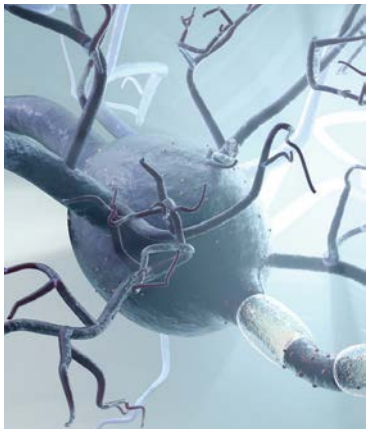
"Join our community and support one another and encourage other families as this is what keeps us going"

STXBP1 Disorders

www.stxdisorders.org
stxbp1@gmail.com

STXBP1 Disorders

Lead the Charge For a Cure



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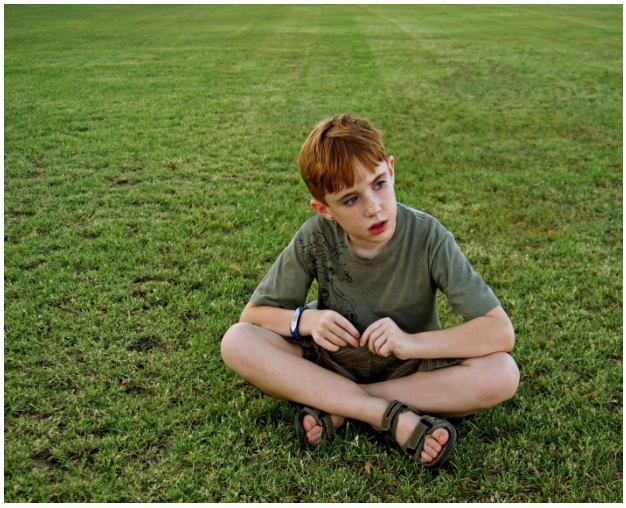
WHAT IS STXBP1 DISORDER?

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.

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 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.

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

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

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).

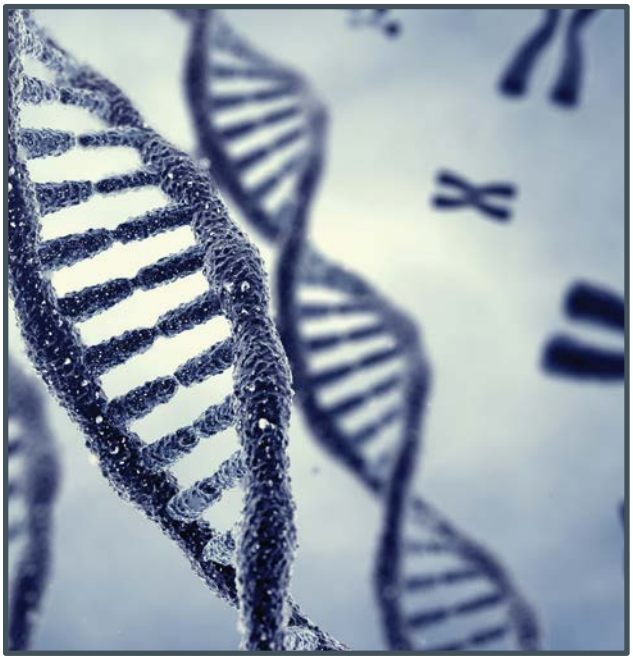
Who are we?

STXBP1 Disorders is part of the *STXBP1* Foundation and is comprised of a group of parents dedicated raising awareness of *STXBP1* Encephalopathy 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.

What are we doing?

We have created a patient registry in order to study the natural history of *STXBP1*. In other words, we want to understand what happens to most children with *STXBP1*. We hope that this registry will one day support clinical trials to "tell us" if a new therapy is working. Currently, we are asking parents to join as we feel that this work will attract more researchers to study *STXBP1*.

"We are asking parents to join as we feel that this work will attract more researchers to study STXBP1"



STXBP1 Disorders