WAYS TO SUPPORT

- Meet the STXBP1 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 STXBP1 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”
**WHAT IS STXB1 DISORDER?**

STXB1 disorder is an autosomal dominant disease, resulting from de novo mutations in the STXB1 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.

**Diagnosis**

STXB1 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 STXB1, or a contiguous gene deletion that includes STXB1 and possibly adjacent genes.

**Incidence**

The disorder occurs in countries, populations, and ethnic groups around the world. The total number of STXB1 patients diagnosed to date based on genetic testing is estimated at 300-400 people worldwide. The estimated incidence of STXB1 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**


**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?**

STXB1 Disorders is part of the STXB1 Foundation and is comprised of a group of parents dedicated to raising awareness of STXB1 Encephalopathy among parents, physicians, scientists, and pharmaceutical innovators. We hope that our work will lead us to better understand the progression of STXB1 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 STXB1. In other words, we want to understand what happens to most children with STXB1. 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 STXB1.

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