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 Searchlight by visiting simonssearchlight.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!

stxbp1disorders.org
info@stxbp1disorders.org

Lead the Charge For a Cure

stxbp1disorders.org
info@stxbp1disorders.org
**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 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.

**Diagnosis**

*STXBP1* diagnosis is made through molecular genetic testing, through a panel test, exome testing or rarely chromosomal microarray analysis.

**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.

**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**