



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!

— Science + L♥ve = Cure —



Join us in finding a cure for this rare genetic disorder



f @stxdisorders
@stxbp1_foundation
@stxbp1disorders
info@stxbp1disorders.org
stxbp1disorders.org

1:30,000
Incidence
rate

1 in 5
display
autistic
features

85%
present
with
epilepsy

López-Rivera et al, 2020, Abramov et al, 2020, Stamberger et al, 2016



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.

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

Brock, D., Rigby, C.S., and Helbig, I. (2021) STXBP1 Disorders. *NORD Rare Disease Database*. <https://rarediseases.org/rare-diseases/stxbp1-disorders/>

López-Rivera JA, Pérez-Palma E, Symonds J, et al. A catalogue of new incidence estimates of monogenic neurodevelopmental disorders caused by de novo variants. *Brain*. 2020;143(4):1099-1105.

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

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 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.

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.

