WAYS TO JOIN AND SUPPORT

- Explore the STXBP1 Foundation website: stxbp1disorders.org
- Participate in natural history studies to accelerate understanding of *STXBP1* and development of better treatment options. Find out more here: stxbp1disorders.org/naturalhistory
- Join our family contact list for parents and primary caregivers: stxbp1disorders.org/contact-list
- Subscribe to the *STXBP1* newsletter: stxbp1disorders.org/newsletter
- Read and contribute to our blog
- Share this brochure with your friends and family
- Follow us on Social Media to learn about the latest news and research: Facebook, Twitter & Instagram and join our Facebook communities.
- Volunteer your time and talents to the STXBP1 Foundation: stxbp1disorders.org/volunteer
- Host a fundraising event: stxbp1disorders.org/fundraise
- Donate to *STXBP1* research: stxbp1disorders.org/ways-to-give
- Join our community, support one another and encourage other families as this is what keeps us going

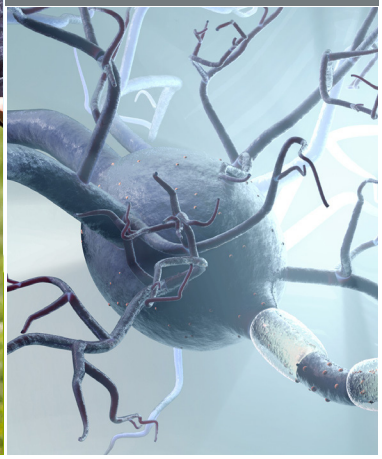
"Join our community, support one another and encourage other families as this is what keeps us going"

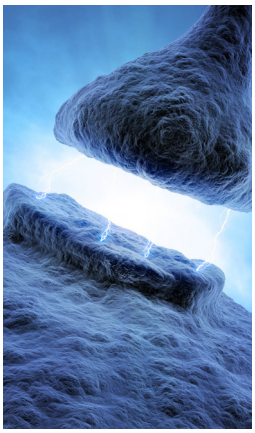
STXBP1 Disorders

stxbp1disorders.org
info@stxbp1disorders.org

STXBP1 Disorders

Lead the Charge For a Cure





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WHAT IS STXBP1-RELATED DISORDERS?

It is a rare neurodevelopmental disorder typically resulting from a *de novo* (new or spontaneous) mutation in the *STXBP1* gene. It is an autosomal dominant gene -- meaning symptoms occur when one of two copies of a gene has a pathogenic change. Pathogenic changes in the *STXBP1* gene cause the cell to not produce enough needed syntaxin-binding protein 1 (STXBP1), an important protein involved in communication between nerve cells.

Signs & Symptoms

There is a broad range of symptoms which may include: early-onset seizures, global developmental delays, intellectual disability, speech/language impairment, muscular hypotonia, movement disorders (such as spasticity, dystonia, ataxia, tremors, or dyskinesia), cortical visual impairment (CVI), and autistic features. Some patients receive other diagnoses such as cerebral palsy or epilepsy syndromes (i.e., Ohtahara syndrome, West Syndrome, Lennox-Gastaut syndrome (LGS)).

An estimated 85% of individuals will have seizures and typically first present in the first year of life, but there have been reports of seizure onset starting later in adolescence. Of those with seizure presentation, about a quarter will not gain seizure control with antiepileptic drugs (AEDs).

Diagnosis

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

Incidence

The disorder occurs in countries, populations, and ethnic groups globally. The total number of *STXBP1* patients diagnosed to date is estimated at 1500 people worldwide. Estimated incidence of *STXBP1* is 1 in 30,000 though true prevalence is unknown. Many cases go undiagnosed or misdiagnosed due to the disorder sharing features commonly seen in other neurodevelopmental disorders. 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.

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. Due to the broad spectrum of symptoms and severity, specific treatment plans are often quite individualized and could benefit from a multidisciplinary team approach. Some specialists involved in the care of those with STXBP1-related disorders may include, but are not limited to: neurologists, neuropsychiatrists, physiatrists, dieticians, gastroenterologists, ophthalmologists, physical and occupational therapists and speech pathologists. Early intervention therapies should be emphasized, including physical therapy, occupational therapy and speech and augmentative communication therapy.

Additionally, seizure management can be a challenge for some and there is no single AED that has been found to be effective for the disorder. While some respond well to a single medication treatment, others require multiple AEDs for adequate seizure control. Ketogenic diet treatment shown variable improvement for seizure management in some individuals.

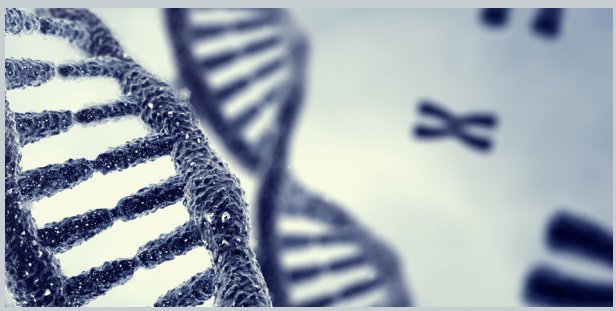
While most patients are nonverbal, some families report their children learning to speak or use sign language to communicate. Patients can benefit from augmentative and alternative communication (AAC) devices.

Who are we?

The STXBP1 Foundation is a parent-led foundation. Our mission is to:

- Create awareness in the disorders associated with STXBP1 mutations
- Fund and drive research to accelerate discovery of a cure
- Provide families with tools to help them understand the disease & how to get involved
- Advocate to improve early detection
- Foster activism to help change policies in favor of orphaned diseases
- Improve the lives of our STXBP1 Family

We hope that our work will lead us to better understand the progression of *STXBP1* and one day lead to a cure.



"Join us as we endeavor to better understand the progression of STXBP1. We hope this work will one day lead to a cure."