WAYS TO SUPPORT

SPREAD AWARENESS
STXBP1 is rare and our families often go for years without the correct diagnosis. Your support in understanding the need for genetic testing and communicating with your colleagues and patients is important to getting our families the information they so desperately need. Additionally:

- Connect families to the STXBP1 Foundation and let them know about our contact registry: stxbp1disorders.org/contact-list
- Keep up with the latest by subscribing to our newsletter: stxbp1disorders.org/newsletter

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.

- Check out STXBP1 funding opportunities: stxbp1disorders.org/applyforagrant
Additionally, there are opportunities for patients and caregivers to contribute to the understanding of this disorder through natural history studies. 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.

- Find more information on natural history studies: stxbp1disorders.org/naturalhistory

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 + Love = Cure ---
WHAT IS STXBP1-RELATED DISORDERS?

It is a rare autosomal dominant neurodevelopmental disorder, resulting from de novo mutations in the STXBP1 gene. Pathogenic mutations are found throughout the gene and cause a haploinsufficiency of the protein Syntaxin-Binding Protein 1 (STXBP1), a part of the SNARE complex. Reduction of the protein STXBP1 impacts the transmission of neurotransmitters into the synapse.

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 undiagnosed or misdiagnosed due to the disorder sharing features commonly seen in other neurodevelopmental disorders. Males and females have equal risk for the disorder.

SIGN & SYMPTOMS

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 seizures, global developmental delays, intellectual disability, feeding difficulties, speech/language impairment, muscular hypotonia, movement disorders (such as spasticity, dystonia, ataxia, tremors, or dyskinesia), and cortical visual impairment (CVI), and autistic features. Some patients receive other diagnoses such as Cerebral Palsy.

An estimated 85% of individuals will have seizures, typically first present in the first year of life, but there have been reports of seizure onset starting later in adolescence. Seizure types can include infantile spasms, generalized tonic-clonic, clonic or tonic seizures, and myoclonic, focal, atonic, and absence seizures. Of those with seizure presentation, about a quarter will not gain seizure control with antiepileptic drugs (AEDs). Associated epilepsy syndromes can include: Ohtahara syndrome, West syndrome, Lennox-Gastaut syndrome, and Dravet syndrome.

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. 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.

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. STXBP1 patients can be found at any age including patients reported in their 60’s.