STXBP1 Disorders

Join us in finding a cure for this rare genetic disease

STXBP1 Disorder is a rare developmental and epilepsy disorder caused by mutations in the STXBP1 gene. The STXBP1 gene is located on chromosome 9. Changes to the STXBP1 gene impact communication between nerves by impairing the release of neurotransmitter in the brain.

5th MOST COMMON diagnosis in patients with genetic testing for epilepsy

Symonds and McTague, 2019

1:30,000 1 in 5 85%

Incidence Rate Display autistic features Present with epilepsy


Key symptoms include epilepsy, global developmental delay, cognitive impairment, autism, and movement disorders. In many children, seizures are the first sign of the condition. In other children, developmental delay may be the initial indication. Currently there are no precision treatments for STXBP1 disorders. Typical treatment for seizures is antiepileptic drugs (AED), and 25% of patients are refractory to AED therapy.

How to Get Involved

Make a Donation  Follow on Social Media
Partner with Us  Volunteer to Help

The STXBP1 Foundation is a 501(c)3 dedicated to raising awareness and finding a cure for STXBP1 disorders. We work with families, physicians, scientists, and pharmaceutical innovators around the world. Help us make a difference.

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