STXBP1 disorders

Science + Love = Cure

JOIN US IN FINDING A CURE FOR THIS RARE GENETIC DISEASE

ABOUT

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.

SYMPTOMS

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 treatmentsfor STXBP1 disorders. Typical treatment for seizures is antiepileptic drugs (AED), and 25% of patients are refractory to AED therapy

5th most common
diagnosis in patients with genetic testing for epilepsy

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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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