JOIN US IN FINDING A CURE FOR THIS RARE GENETIC DISEASE

ABOUT
STXB1P1 Disorder is a rare developmental and epilepsy disorder caused by mutations in the STXB1P1 gene. The STXB1P1 gene is located on chromosome 9. Changes to the STXB1P1 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 treatments for STXB1P1 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 STXB1P1 Foundation is a 501(c)3 dedicated to raising awareness and finding a cure for STXB1P1 disorders. We work with families, physicians, scientists, and pharmaceutical innovators around the world. Help us make a difference.

MAKE A DONATION  ●  PARTNER WITH US  ●  FOLLOW US ON SOCIAL MEDIA  ●  VOLUNTEER TO HELP

1:30,000 incidence rate  1 in 5 display autistic features  85% present with epilepsy