September is STXBP1 Awareness Month!

Get ready for all things STXBP1. This is our third year of dedicating the entire month of September to STXBP1 Disorders with ongoing communications and activities.

Why September? September is the 9th month and STXBP1 gene is on the 9th chromosome.

Join in all month long and we’ll work together to create more awareness for this rare genetic disorder, keep up with the very latest research, and bring the community together through our Annual Move to Cure Event ... virtually.

STXBP1disorders.org
#STXBP1
Did you know...

Q. What is STXBP1?

A. STXBP1 is a gene that is involved with brain development and neurotransmitter signaling. It is also known as Munc18-1. Changes in the STXBP1 gene (mutations) can result in epilepsy, intellectual disability, developmental delay, movement disorders, and other difficulties.

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Did you know...

Q. How are STXBP1-related disorders diagnosed?

A. In a child with features of STXBP1-related disorders such as early onset epilepsy, a doctor may order a gene panel or microarray test that identifies mutations, or variants, in the STXBP1 gene. STXBP1 variants may also be diagnosed using newer genetic tests called whole exome and whole genome sequencing.

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Did you know...

Q. Is there a cure for STXBP1-related disorders?

A. There is currently no cure for STXBP1-related disorders. However, physicians can help manage some of the symptoms of STXBP1, such as treating seizures if they occur.

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Q. Is there a standard treatment for disorders caused by STXBP1?

A. There is no standard treatment at this time. Commonly used anti-epileptic drugs (AEDs) include phenobarbital, valproic acid and vigabatrin. 25% of patients do not respond to AEDs. Many patients also benefit from physical, occupational, feeding and speech therapy.

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Did you know...

Among patients with epilepsy who had genetic testing, STXBP1 is the 5th most common genetic diagnosis.


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Did you know...

Q. Will my child continue to have seizures after treatment?

A. Most children can have their seizures controlled with medications. However, some children will have epilepsy that is very difficult to control, even with multiple medications.

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Did you know...

Q. Did I cause this gene mutation in some way?

A. No. STXBP1 mutations occur completely by chance, and are not affected by anything in the environment. Dietary changes, medications, vitamins, or lifestyle factors cannot cause (or prevent) gene mutations.

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Did you know...

Q. What other problems do children with STXBP1 mutations have?

A. Patients have a wide range of symptoms. Most children have some degree of cognitive impairment, varying from mild to profound. Movement disorders and autistic features are common. Many children have trouble with feeding and swallowing. Most patients are nonverbal, though some speak and/or sign.

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Did you know...

85% of patients present with epilepsy. All patients present with developmental delay or intellectual disability.


STXB1P1disorders.org
#STXB1P1
Did you know...

1 : 30,000 incidence rate


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