

STXBP1 gene

syntaxin binding protein 1

Normal Function

The *STXBP1* gene provides instructions for making syntaxin-binding protein 1. In nerve cells (neurons), this protein helps regulate the release of chemical messengers called neurotransmitters from compartments known as synaptic vesicles. The release of neurotransmitters relays signals between neurons and is critical for normal brain function.

To release its neurotransmitters, a synaptic vesicle must join (fuse) with the outer membrane of the neuron. The syntaxin-binding protein 1 regulates the formation of a group (complex) of proteins that allows vesicle fusion.

Syntaxin-binding protein 1 may also have a role in the positioning and growth of neurons during brain development. Proper localization of neurons is important for normal brain formation and function.

Health Conditions Related to Genetic Changes

STXBP1 encephalopathy

More than 150 mutations in the *STXBP1* gene have been found to cause *STXBP1* encephalopathy. This condition is characterized by abnormal brain function (encephalopathy) and intellectual disability. Most affected individuals also have recurrent seizures (epilepsy) that begin in infancy. The *STXBP1* gene mutations can alter the structure of syntaxin-binding protein 1, result in an abnormally short protein, or add or delete small sections of the protein.

The gene mutations that cause *STXBP1* encephalopathy reduce the amount of functional syntaxin-binding protein 1 produced from the gene. A shortage of this protein impairs the formation of the protein complex that allows vesicle fusion and the release of neurotransmitters from neurons. A change in neurotransmitter levels can lead to uncontrolled activation (excitation) of neurons, which causes seizures. Researchers suspect that a shortage of syntaxin-binding protein 1 also impairs neuron development in certain regions of the brain, which could underlie abnormal brain function and other neurological problems in people with *STXBP1* encephalopathy.

Lennox-Gastaut syndrome

MedlinePlus Genetics provides information about Lennox-Gastaut syndrome

Other Names for This Gene

- hUNC18
- MUNC18-1
- N-Sec1
- neuronal SEC1
- NSEC1
- RBSEC1
- unc-18A
- UNC18
- unc18-1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of STXBP1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6812\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6812[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28STXBP1%5BTIAB%5D%29+OR+%28syntaxin+binding+protein+1%5BTIAB%5D%29+OR+%28MUNC18-1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D%29%29%29>)

Catalog of Genes and Diseases from OMIM

- SYNTAXIN-BINDING PROTEIN 1; STXBP1 (<https://omim.org/entry/602926>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/6812>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=STXBP1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=STXBP1[gene]))

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Genomic Location

The *STXBP1* gene is found on chromosome 9 (<https://medlineplus.gov/genetics/chromosome/9/>).

Last updated December 2, 2020