

What causes disorders of *STXBP1*?

STXBP1 is a gene which is important for normal brain function. It plays a vital role in the release of chemicals in the brain (neurotransmitters) which the brain cells use to communicate with each other. A disruption or the absence of this gene results in abnormalities in the function of the brain leading to seizures, developmental delay, learning difficulties and behaviour problems.

Why did this happen?

When children are conceived your genetic material is copied in the egg and sperm that makes a new child. The biological copying method is not perfect and occasionally random rare changes occur in the genetic code of children that are not seen in the DNA of their parents. Disorders of *STXBP1* occur when one of these random, rare changes affects the *STXBP1* gene on chromosome 9. This happens naturally in plants and animals and is not due to your lifestyle or anything you did to cause a change in the *STXBP1* gene. In most families the DNA change in *STXBP1* occurs out of the blue. This is what you may hear a geneticist referring to as a de novo change.

Can it happen again?

The possibility of having another child affected by a rare gene disorder depends on the genetic code of the parents. For *STXBP1* disorders where parents do not carry the mutation the chance of having another affected child is very low. If the genetic analysis of the parents of a child with an *STXBP1* disorder shows they carry the same variant, the chance of it happening again is much higher. Each family situation is different and a clinical geneticist can give you specific advice for your family.

Families say ...

“STX kids are known for their happy and easy going demeanours.”

“STX is a disease that can show up in many different ways. No child has the same manifestation, though there are some similarities. It is hard to know what your child will be able to do and what limitations they will have. There is a great need for families facing this diagnosis to have support dealing with what it means to their family and figuring out how to navigate the medical system to get the help and resources they will need.”

Inform Network Support

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Facebook Parents of Kids with STXBP1
www.facebook.com/groups/123917951055617



Unique lists external message boards and websites in order to be helpful to families looking for information and support. This does not imply that we endorse their content or have any responsibility for it.

This guide is not a substitute for personal medical advice. Families should consult a medically qualified clinician in all matters relating to genetic diagnosis, management and health. Information on genetic changes is a very fast-moving field and while the information in this guide is believed to be the best available at the time of publication, some facts may later change. Unique does its best to keep abreast of changing information and to review its published guides as needed. This guide was written by Dr Andreas Brunklaus, Consultant Paediatric Neurologist, Royal Hospital for Sick Children, Glasgow, Scotland, UK with contributions from representatives of a parent group. 2015 Version 1 [PM]

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Disorders of *STXBP1* rarechromo.org

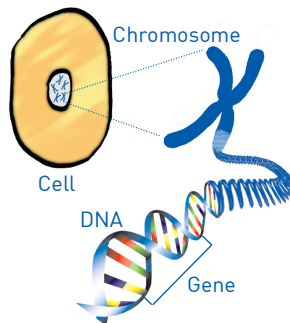


What are the disorders of *STXBP1*?

STXBP1 is a gene on chromosome 9 which has been found to be linked with severe epilepsy in very young babies. More recently changes (mutations) in the *STXBP1* gene have also been found in children without a seizure disorder, but with learning difficulties. The *STXBP1* gene is found on chromosome 9 in band 9q34.11 at base pairs 130374544 to 130457460 [hg19 genome assembly].

STXBP1 also causes movement disorders including unsteadiness [ataxia], abnormal muscle function [dystonia], tremors and low muscle tone [hypotonia].

In a small number of cases children may have Ohtahara syndrome, also known as early onset epileptic encephalopathy with suppression-bursts or infantile spasms, also known as West syndrome.



Most people with a disorder of *STXBP1* have:

- Intellectual disability or developmental delay
- Seizure disorder
- Movement disorder

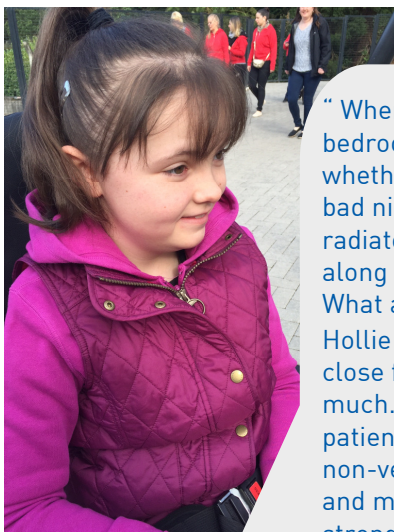
Can this be cured?

There is no cure for this disorder as the effects of the genetic change took place during your baby's formation and development. However, knowing the diagnosis means that appropriate monitoring and treatment can be put in place for your child.

Was the diagnosis helpful?

- Having a diagnosis allowed us some closure in the sense that we didn't have to keep searching for a reason why he was having the symptoms. We also were able to connect with other families who had the same diagnosis and that provided much needed support and understanding.

" He is extremely sweet and loving. He loves to smooch us and he is delightfully happy. He has changed our lives in every possible way. Some things are much harder, yet he has brought a love that we have never known and could not imagine before we had a child." 3 years



" When I walk into her bedroom in the morning, whether it has been a good or bad night, the room is radiated by a beautiful smile along with a High Five. Wow! What a great start to the day! Hollie has made us a very close family - we love her so much. We have all gained patience, understanding her non-verbal communication and most of all we stand strong to fight her corner every inch of the way. " 11 years

Related medical conditions

Ohtahara syndrome and seizures

Ohtahara syndrome is a very debilitating neurological disorder. Seizures often start in the first weeks of life and are commonly very severe. They frequently occur daily and are difficult to control with medication. Developmental delay is typically severe, and, sadly, not all babies survive.

West syndrome and seizures

In West syndrome brief stiffening seizures (spasms) start between 4-7 months of age, and these may be controlled with medication. Some children may remain seizure-free. Children with West syndrome can develop mild to severe intellectual disability later in life.

Development

■ Sitting, crawling, on the move
STXBP1 disorders commonly affect movement and mobility. A few children may walk once put on their feet, while others need full adult support to do so. Balance, co-ordination and muscle tone problems contribute to these mobility issues.

■ Learning
Most children need substantial support with their learning and are likely to attend a special school where the right support can be given and non-academic and daily living skills are focused on. Supervision is likely to be needed even for adults.

■ Behaviour
As with many children who have severe epilepsy behaviour is often influenced by the frequency of seizures and effects of medication. Autistic traits have been observed in a number of children.

■ Speech
Children may not develop speech. Instead they communicate by other means, such as gestures and vocal noises.

■ Using their hands
Children may have repetitive hand movements.

Management recommendations

■ Children with an *STXBP1* mutation should be under the care of a multidisciplinary team. Expert advice regarding seizure management is available from an epilepsy specialist as well as an epilepsy nurse.

■ Paediatricians and/or community paediatricians should oversee care so that development, growth and behaviour can be monitored and the best help in the form of physiotherapy, occupational, speech and behavioural therapies (including medications) can be given early if needed.

Was the diagnosis helpful?

- Yes and no. Yes - we know the reason for her difficulties, and no, as it is rare and there is not enough information.