Why did this happen?

There is no known cause for SOX11 variants or 2p25.2 deletions. Nothing you did, or did not do has caused this.

Can it happen again?

Provided that neither parent carries the same genetic change as their child the chance of having another affected child is extremely low - less than one per cent. The reason for this residual risk is something called 'gonadal mosaicism'. This means that people can, very rarely, carry a genetic variant/change in their sperm or eggs but not in the other cells of their body. This would mean that the genetic variant would not be detectable on a blood or saliva sample. Specific advice should be sought from your clinical genetics team.

How many people have SOX11 syndrome?

SOX11 syndrome is a very rare condition. To date only seven children with spelling mistakes of the SOX11 gene have been reported and seven people with deletions of the SOX11 gene. However, with increasing use of 'next generation' genetic diagnostic technology we will identify more people affected by this condition. This will help us provide more useful information to families.

What is 2p25.2 deletion syndrome and how is it related to SOX11 syndrome?

A well recognized cause of neurodevelopmental conditions is deletion (loss) of chromosome segments. This can result in the loss of many different adjacent genes (instructions). The SOX11 gene is found at chromosome 2p25.2. Deletions affecting this region of chromosome cause a neurodevelopmental condition with features of SOX11 syndrome. Generally speaking people with 2p25.2 deletions have similar features to people with SOX11 'spelling mistakes'. However, some of the people with this deletion have been more severely affected due to loss of genes which sit next to the SOX11 gene.

Families say ...

" Ava is currently 2 years and 11 months. She has a very laid back nature, and is extremely sociable. She has a good imagination, and is very playful. Although slightly delayed, her speech is vastly improving, and Ava can now communicate her needs effectively. She is seeing a speech and language therapist who is investigating her continuous dribbling, with the possible cause being muscle control. She is currently attending nursery and her EYFS (early years foundation stage) scores are within the typically developing range. Potty training is still an ongoing challenge, with occasional mistakes of one accident per day. However Ava's ability to entertain through singing and dancing is a constant joy, heightened by her love for dressing up! "

Inform Network Support

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Understanding chromosome disorders Unique

SOX11 syndrome

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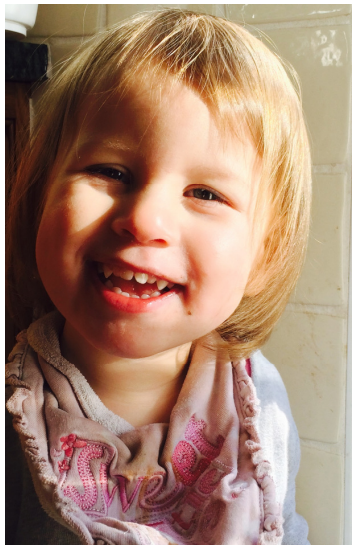

What is SOX11 syndrome?

SOX11 syndrome is a recently discovered condition. Clinical geneticists would refer to it as a neurodevelopmental disorder. For clarity the term SOX11 syndrome will be used in this leaflet.

SOX11 syndrome happens when one of a person's two copies of the SOX11 gene does not function as it should. Genes are the instructions which tell our bodies how to develop and function. Genes are made of DNA and packaged into thread-like structures called chromosomes, which are found in almost every cell of our body. The SOX11 gene is found on chromosome 2 and is thought to give an important instruction for brain development. This is why alterations in the SOX11 gene are mainly associated with a neurodevelopmental disorder. These alterations can either be spelling mistakes in the genetic code of SOX11 or loss of one copy of the SOX11 gene (deletion).

What is 2p25.2 deletion syndrome and how is it related to SOX11 syndrome?

A well recognized cause of neurodevelopmental conditions is deletion (loss) of chromosome segments. This can result in the loss of many different adjacent genes (instructions). The SOX11 gene is found on chromosome 2 in band 2p25.2 at base pairs 5696666-5701384 [hg19 genome assembly]. Deletions affecting this region of chromosome 2 cause a neurodevelopmental condition with features of SOX11 syndrome. Generally speaking people with 2p25.2 deletions have similar features to people with SOX11 'spelling mistakes'. However, some of the people with this deletion have been more severely affected due to loss of genes which sit next to the SOX11 gene.

Why did this happen?

There is no known cause for SOX11 variants or 2p25.2 deletions. Nothing you did or did not do has caused this.

Can this be cured?

There is no cure as the effects of the genetic change took place during development in the womb. However, knowing the diagnosis means that appropriate monitoring and treatment can be put in place.

Most common features

Everyone with SOX11 syndrome has developmental delay or learning difficulties. Other typical features include:

- Small head
- Marked speech delay
- Poor feeding as a young baby
- Seizures
- Inward curvature of the 5th [little] finger

None of these features is specific for SOX11 syndrome. Diagnosis must be made with a specific genetic test.

Medical concerns

Seizures/epilepsy

Some children with SOX11 syndrome have experienced seizures, including generalised and absence seizures. These have responded to standard drug treatment for epilepsy.

Skeleton

Some children with SOX11 syndrome have scoliosis (a sideways curvature to the spine). This has generally been mild and not required treatment. Most children with SOX11 syndrome have inwards curving 5th fingers with small fingernails on their 5th fingers. This does not affect hand function.

Eyesight

Some children with SOX11 syndrome have short or long sight or a squint. One child with a SOX11 deletion had one eye which was smaller than it should have been. It is not clear if this was just chance or part of SOX11 syndrome.

Feeding difficulties

Many newborn babies with SOX11 had feeding difficulties requiring temporary nasogastric feeding. No child has required long term nasogastric tube feeding.

Neuroimaging

One child with SOX11 syndrome had a slight reduction in the size of a part of the brain called the cerebellum. One child with SOX11 syndrome had a slight reduction in the size of the electrical wiring connecting the 2 halves of the brain (corpus callosum). Changes on brain scans can be caused by SOX11, but we do not have enough information yet to be certain on the precise types of brain changes which can happen in SOX11 syndrome.

Development

Growth

Children with SOX11 syndrome are generally of average birth weight. Height in childhood lies within the average range. Head circumference is reduced, but not so much as to be noticeable to a casual observer.

Sitting, walking, moving

All children had some degree of delayed motor development. However, all achieved independent walking.

Speech

All but one child studied so far had delay in speaking (first words spoken from 18-36 months). At the age of 12 one child had no speech.

Learning

All children we know of have required some help with learning at school age.

Behaviour

Some children with SOX11 syndrome have been diagnosed as autistic. Children with SOX11 syndrome have no characteristic alterations to their behaviour.

Management recommendations

- Feeding management if necessary (for example nasogastric tube feeding as a baby)
- EEG (measurement of the brain's electrical activity) if seizures are suspected
- Eye check
- Brain imaging with MRI (for example if a child has seizures or limb weakness)
- Follow up by a developmental paediatrician
- Physiotherapy and speech and language therapy as needed.

Age 2