What causes FOXP2 disorder?

FOXP2 is a gene which has important functions for the development of the brain. A disruption or absence of one gene copy results in difficulties in the acquisition of speech and language.

Why did this happen?

In many families, the genetic change affecting FOXP2 happens out of the blue (de novo). When children are conceived, the genetic material of the parents is copied in the egg and sperm that makes a new child. The biological copying machine 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. This happens naturally in plants and animals and is not due to your lifestyle or anything you did. In some families, one parent has the same FOXP2 change and has transmitted this to the child.

Can it happen again?

The probability of having another child affected by a rare gene disorder depends on the genetic code of the parents. For FOXP2 defects where parents do not carry the mutation, the chance of having another affected child is very low. If genetic analyses of the parents show that one of them carries 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.

Management:

General paediatricians should oversee care, so that development and behaviour can be monitored and the best therapies can be given early. Speech sound development and language abilities should be assessed by a speech therapist to determine the nature of a child's communication strengths and challenges and how to best support their needs and development. Every child's communication profile is different and no single treatment exists. Instead, treatment and management approaches will depend on the type and severity of a child's difficulties. This may include intensive speech sound therapy and/or augmentative and alternative communication (AAC) options tailored to an individual child's needs.

Other health professionals such as clinical psychologists (for learning difficulties), physiotherapists (for whole body movement challenges such as learning to ride a bike), occupational therapists (for fine motor challenges such as writing) can provide strategies to help with other specific problems a child may have.

Families say ...

"When you have a child that has difficulties in everyday life it makes them that little bit more special!"

"He is an extremely happy boy and interacts with everyone and has done since he was a newborn.
The major area of his development that has been affected is his speech and language. He understands everything we tell him; however, he cannot express himself with speech which, at times, frustrates him a lot. At 3½ years he has a couple of words he can clearly say ('mum' and 'juice', 'choo choo' for train) and a couple of words he can try to say (e.g. tree he pronounces as 'ee' and car as 'ar'). In general he communicates via 'babble' talk, pointing and taking someone's hand to show them. We also try to use a handful of Makaton signs."

Inform Network Support

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Gene reviews article:
https://www.ncbi.nlm.nih.gov/books/NBK368474/

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Unique
Understanding Chromosome & Gene Disorders

What is FOXP2 related speech & language disorder?

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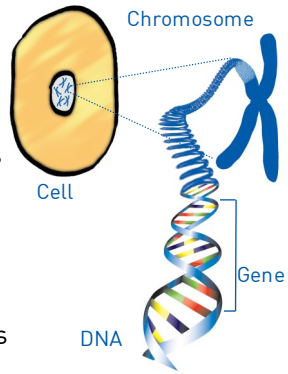


FOXP2 is a gene on chromosome 7q31.1, which was first found to be linked with speech and language disorders in 2001. Affected individuals were noted to have childhood apraxia of speech (CAS), which is a speech disorder that results in difficulties in the planning and production of sounds and words. Some also have problems with reading and spelling or learning difficulties. Some individuals have a disruption of the FOXP2 gene only, and some have a more complex genetic alteration that involves additional genes (e.g., FOXP2 deletion or duplication). Hence the associated genetic conditions are known as 'FOXP2-only' and 'FOXP2-plus', respectively. The genetic alteration determines whether speech and language problems are seen in relative isolation, or whether more general developmental issues across a range of areas are likely to be present as well.

Can it be cured?

There is no cure for FOXP2 defects, as the effects of the genetic change took place during your baby's development. However, knowing this diagnosis means that appropriate monitoring and therapies (e.g. speech therapy) can be put in place for your child.

Families say ...

"She has completed our family. She has shown us all how to stay positive and be proud of everything you do no matter how small. She makes me smile everyday! She is a delightful child and never lets her condition get her down.

We noticed when she was around 1 1/2—2 years old that she was not speaking as much as her brother and sister did at that age. She is now 7 years old and able to put three or four words together.

She is able to follow commands without any problem. We try to take the time to listen to her and not speak for her, even if it is time-consuming."

Most people have:

- Delayed speech development
- Motor speech disorder (childhood apraxia of speech)
- Expressive and/or receptive language difficulties
- Reading and spelling difficulties
- Normal or low average IQ
- Normal organ functions

Children with the FOXP2-plus genetic condition, involving a deletion or duplication disrupting genes that neighbour FOXP2, may have associated learning, behavioural or social skill challenges.

Medical concerns

Childhood apraxia of speech

Young children with FOXP2-only gene changes often have delayed speech development. From around age three years, when they have developed enough speech for a thorough speech therapy assessment, most children are diagnosed with a speech disorder, called childhood apraxia of speech (CAS). CAS is a motor speech planning problem affecting a person's ability to accurately sequence sounds into syllables, syllables into words and words into sentences with the correct timing and rhythm. As a result speech can be very difficult to understand, particularly in the early childhood and primary school years. Speech development and intelligibility does improve over time, but ongoing management and evaluation is recommended in most cases.

Learning

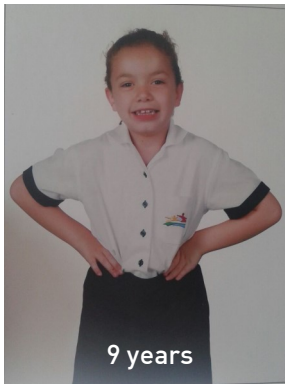
Many children have normal or low average IQ, and nonverbal IQ is often higher than verbal functions. People with FOXP2 defects often have reading and spelling difficulties.


4 years

Healthy organs

No malformations of organs (for example heart, kidneys, brain) are associated with FOXP2 defects.

Development


9 years

Growth

Almost all babies are average in weight at birth and continue to grow along their centile lines.

Feeding

Some babies have early feeding difficulties due to their poor oral motor functions. Dribbling can be a problem for many children but this can be helped by medicines to reduce the saliva.

Sitting, moving and walking

Gross motor development is normal in many children with only FOXP2 defects. Some children may have fine motor difficulties (e.g. buttoning clothes, tying shoelaces). Fine or gross motor problems are reported as relatively mild compared to the marked speech production deficits. If it is noted that a child is late in becoming mobile or having problems with fine motor skills, physiotherapy or occupational therapy can be useful.

Speech

Speech development is the major concern in FOXP2 defects, with the primary speech diagnosis of childhood apraxia of speech. Some children may also have dysarthria. Most children need considerable speech therapy and support.

Language

Some individuals may experience problems in expressive (using spoken language) and/or receptive (understanding spoken language) domains.

Learning

Nonverbal (performance) IQ is typically normal or low average. Some children need support with their learning (for example reading and spelling abilities) and may need to attend a special school where the right support can be given.

Behaviour

On the whole, children are sociable and have a friendly character. Autistic traits have only been noted infrequently and only in children with the FOXP2-plus condition involving neighbouring genes to FOXP2.