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.

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

Rare Chromosome Disorder Support Group,
The Stables, Station Road West, Oxted, Surrey RH8 9EE, UK
Tel/Fax: +44(0)1883 723356
info@rarechromo.org | www.rarechromo.org

This guide was written by Dr Christiane Zweier and Dr Miriam Reuter, Institute of Human Genetics, FAU, Erlangen, Germany.
2017 Version 1 (PM/CA)

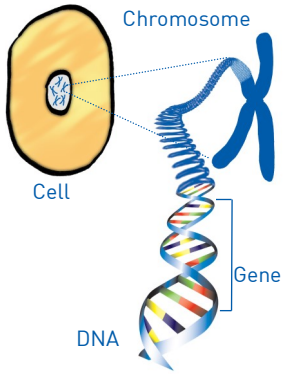
Rare Chromosome Disorder Support Group
Registered in England and Wales
Charity Number 1110661
Company Number 5460413

Understanding Chromosome & Gene Disorders

What is FOXP2 related speech & language disorder?

rarechromo.org

What is FOXP2 related speech & language disorder?



FOXP2 is a gene on chromosome 7q31.1, which was first found to be linked with speech and language disorders in 2001. Some individuals have a defect only of FOXP2, and some have a more complex genetic alteration that involves additional genes. Affected individuals were noted to have difficulties in the planning and production of sounds and words. Some also have problems with reading and spelling or learning difficulties.

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 developmental and speech therapies 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 and language disorder
- Reading and spelling problems
- Normal or low average IQ
- Normal organ functions

Medical concerns

Childhood apraxia of speech

Young children often have delayed speech development. From age three years, many children develop a typical speech disorder, called childhood apraxia of speech. It is characterized by oral motor difficulties, with problems turning sounds into syllables, and syllables into words. Understanding of language can also be affected.

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.

Healthy organs

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

Management:

General paediatricians should oversee care, so that development and behaviour can be monitored and the best therapies can be given early.
Speech sounds should be assessed by an ear, nose, and throat specialist.
Speech therapists, clinical psychologists and physiotherapists can provide strategies to help with the specific problems your child may have.

Development

- 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

Fine and gross motor development is normal in many children with only FOXP2 defects. If it is noted that a child is late in becoming mobile, physiotherapy can be useful.

- Speech

Speech development is the major concern in FOXP2 defects, and most children need considerable support.

- Cognitive functions

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.