Why did this happen?

In most families, the genetic change affecting the FOXP1 gene happens out of the blue (this is known as *de novo*). When children are conceived, the genetic material of the parents is copied in the egg and sperm which makes a child. The biological copying method is not perfect and random rare changes occur in the genetic code of a baby, that are not seen in parents' DNA. This happens naturally in everyone and is not due to anything parents did or did not do; it's only when changes affect an important gene that symptoms are seen. (Two parents have been found to have an unusual chromosomal rearrangement that caused a FOXP1 gene change in their 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 FOXP1 variants where parents do not carry the genetic change, the chance of having another affected child is very low (less than 1%). If genetic analyses of the parents show that one of them carries a disruption of the FOXP1 gene, the chance of the child having an altered FOXP1 gene copy is much higher (about 50%). Each family situation is different and a clinical geneticist or genetic counsellor can give you specific advice for your family.

Management:

General paediatricians should oversee care, so that development and behaviour can be monitored and ensure early access to therapy.

Individuals with FOXP1 syndrome should be referred for comprehensive neuropsychological evaluations and have a neurology and/or developmental paediatrics referral to assess a child's motor skills. Consultations should be followed by appropriate referrals to paediatric physiatry and orthopaedics.

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 therapy to treat speech disorders; augmentative and alternative communication (AAC) options should be considered.

Physiotherapists and occupational therapists can also provide support. Individuals with FOXP1 syndrome should be referred for hearing and vision evaluations.

Families say ...

"Helping our daughter to walk and talk was challenging, but the difficult behaviours and the obsessions we deal with today takes parenting to a whole new level. Thank goodness for her sense of humour, that get us through our dark days."
- Daughter aged 15.

"As a child, he was an absolute joy. Happy, enthusiastic and just very lovely. He was delayed in all his development but would happily have a go at anything he was asked to do. He attended a primary school nursery and infant school as well as a special school in a split placement. He loved being around other children. As he's got older, he can be more challenging and has autism with all its complexities. However, he has developed a lovely sense of humour and is charmingly sociable. He likes to have a full life and embraces all actives available ... except dancing and drama! He lives in his own house and is supported by a 24/7/52 support package. Life has not been kind to him but he has the best life he could have." - Son aged 38.

Inform Network Support

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Websites and Facebook groups:

www.foxp1.org/
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http://kindbeter.nl/ (Dutch)

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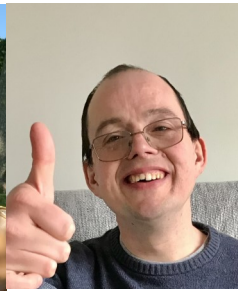
Understanding Chromosome & Gene Disorders

FOXP1 syndrome



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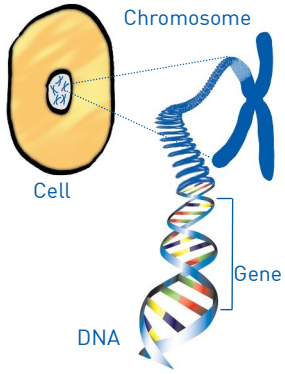
What is FOXP1 syndrome?

FOXP1 syndrome is a rare genetic syndrome associated with speech and language difficulties, global developmental delay and mild to moderate intellectual disability. People with FOXP1 syndrome can also have autism spectrum disorder or autistic features, motor delays and low muscle tone, together with other features which vary between individuals.



So far over 100 people with FOXP1 syndrome have been reported in the medical literature. However, many more are known to have this diagnosis and there are likely to be many people who have not yet been diagnosed.

What causes FOXP1 syndrome?

FOXP1 is the name of a gene, the gene is located on chromosome 3 in a region known as 3p14.1. Each person has two copies of chromosome 3 and so usually has two copies of the FOXP1 gene. FOXP1 syndrome is caused by a change to one of these two copies. The genetic change is known as autosomal dominant which means symptoms are seen when only one copy of the gene is affected.

Can it be cured?

There is no cure for FOXP1 syndrome, since the genetic change effects a baby's early development. However, knowing this diagnosis means that appropriate monitoring and therapies can be put in place.

Most people with FOXP1 syndrome have:

- Global developmental delay (GDD)
- Mild to moderate intellectual disability (ID)
- A language disorder (expressive skills are greater than receptive skills)
- A speech disorder (speech apraxia, dysarthria)
- Autism Spectrum Disorder (ASD) or autistic features
- Low muscle tone (hypotonia)
- Gross and fine motor delays
- Behavioural and mental health difficulties

Some people with FOXP1 syndrome have:

- Brain and heart anomalies
- Seizures
- Hearing and vision difficulties

> "She is a very loving and caring little girl. She has a beautiful smile and a great sense of humour! She loves to sing and dance along to her favourite films and TV shows. She has taught us to see the world differently, to be patient, to slow down and to appreciate & celebrate every small achievement." - Daughter aged 10.

Medical concerns

Hypotonia

About a third of children with FOXP1 syndrome reported in the medical literature to date (2021) have been diagnosed as having low muscle tone early in life. Facial hypotonia can contribute to speech and feeding difficulties. Some children have also been recorded to have muscle spasms and hypertonia (high muscle tone).

Brain

About half of children assessed so far have been found to have unusual findings during a brain MRI but no consistent structural brain change has been identified.

Seizures

Less than 10% of children reported so far experience seizures.

Heart and Lungs

Almost a third of individuals reported to date have some form of heart anomaly. Some require medical or surgical intervention. Some children also experience recurrent chest infections.

Urogenital

Some children have been reported as having genitourinary malformations and others have difficulties with incontinence.

Vision

About half of individuals have been reported as having some form of eye or sight anomaly, such as being longsighted, short-sighted, having poor vision in one eye or having a squint.

Hearing

Some individuals have also been reported as having hearing loss or recurrent ear infections. Hearing loss severity differs amongst children.

Facial features

Some children with FOXP1 syndrome may have subtle facial features such as, a prominent forehead and short nose with a broad tip, base or bridge. Children may also have eyes that slant downwards, droop or are widely spaced.

Development

Feeding

Some babies have early feeding difficulties due to their poor oral motor functions.

Motor development

Gross motor development (e.g., walking) is affected in many children with FOXP1 syndrome. Most children also have fine motor difficulties (e.g., holding a pencil). Physiotherapists can provide support for children with gross motor difficulty and occupational therapists can support children with fine motor difficulties.

Speech

Speech, or the sounds we make when we talk, is often a main area of developmental concern in FOXP1 syndrome. All children reported in the literature to date have had speech difficulties. Dysarthria, a speech disorder affecting the precision of speech is the most common speech difficulty, seen in unclear speech sounds, unusual stress patterns and poor voice quality. Other speech disorders may also be seen including features of apraxia of speech (difficulty coordinating sounds affecting the accuracy of speech) and phonological disorder (substituting sounds). Most children benefit from support from a speech therapist.

Language

Most individuals will have trouble using and understanding words and sentences. A child's ability to use language (e.g., expressive language skills) is often stronger than their ability to understand (e.g., receptive language skills). Whilst it is common to assume that receptive language abilities are better because of speech difficulties, research has shown this is not the case for FOXP1 syndrome. Rather, children often have better expressive vocabulary and sentence production than understanding in these areas. Most children are late talkers. Some children may require other systems to communicate, such as picture systems (AAC) or sign language.

Learning

Most children are diagnosed with mild to moderate intellectual disability (nonverbal IQ). Children will need support with their learning (for example, with literacy skills). Many children may benefit from attending a special school where they can receive more support.

Behaviour

Most children are very social. Autistic spectrum disorder has been diagnosed in many children. Other diagnoses include ADHD, aggression, obsessive-compulsive traits, sensory difficulties, mood disorders and anxiety.