Translational Centre for Speech Disorders
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FOXP2-related speech and language disorder (*FOXP2*-SLD)

Fact sheet

What is *FOXP2-related speech and language disorder*?

FOXP2 is a gene on chromosome 7q.31.1. *FOXP2* is a transcriptor protein which controls the activity of other genes.[1] *FOXP2* is important for brain development (pre and post birth) and growth of nerve cells. *FOXP2* protein is important in the transmission of signals between the brain and the nerve cells and plays an important role in synaptic plasticity[1].

FOXP2-related speech and language disorder (*FOXP2*-SLD) occurs when there is a pathogenic change or variation (like a spelling mistake, called a variant) in the *FOXP2* gene, meaning the gene does not function as efficiently. A pathogenic *FOXP2* change results in altered development.

Speech and Language

The terms 'speech' and 'language' are often used interchangeably; yet, they are categorised differently by a speech pathologist, with has implications for therapy:

Speech is focused on speech sounds. This includes sound accuracy, articulation, voicing, resonance (e.g., nasality), and prosody (e.g., stress and rhythm).

Language involves the understanding and use of words (vocabulary) and sentences (grammar).

FOXP2-plus related speech and language disorder is a distinct but related condition to *FOXP2*-SLD. The *FOXP2*-plus condition occurs when there is a deletion or duplication of chromosome 7q31.1 that involves not only *FOXP2,* but also neighbouring genes.

The type of pathogenic *FOXP2* variant that the individual has will determine whether only speech and language is impacted (*FOXP2*-related speech and language disorder) or whether the individuals will experience broader (global) developmental delays or behavioural difficulties (*FOXP2*-plus related speech and language disorder).[3] Childhood Apraxia of Speech (CAS) is a key feature in individuals with *FOXP2*-SLD. Approximately 1 in 3 children with CAS have a genetic cause for their CAS. However, *FOXP2*-SLD is very rare, even amongst children with CAS[3].

The Information provided on this webpage pertains to *FOXP2*-SLD (individuals with variants affecting only the *FOXP2* gene) and not larger deletions (7q31.1 deletions, as seen in *FOXP2*-plus related speech and language disorder).

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What are the associated health and medical conditions seen in *FOXP2*-SLD?

Individuals with *FOXP2*-SLD typically experience CAS, verbal cognition (intelligence quotient) and expressive and receptive language challenges, and fine motor skill difficulties, such as doing up small buttons or holding a pencil.[3,6] Some individuals have mild intellectual disability, whilst others do not have an intellectual disability.[4] Literacy skills, such as reading and writing may also be challenging for individuals with *FOXP2*-SLD.[3,6] Almost half of individuals with *FOXP2*-SLD have sleep disturbances.

A small number of individuals are autistic.[6] Many individuals have hypotonia (low muscle tone).[3] Approximately half of individuals report gross motor challenges, such as learning to walk, in early childhood. Approximately 1/4 of individuals have both fine and gross motor challenges.[6]

By contrast, individuals with *FOXP2*-plus speech and language disorder (with large 7q.31.1 deletions encompassing the *FOXP2* gene) commonly experience more cognitive and learning challenges than individuals with *FOXP2*-SLD. these challenges can include intellectual disability, global developmental delay, and autistic features.[3]

In infancy, many individuals with *FOXP2*-SLD do not babble.[3] Young children with *FOXP2*-SLD may present with fine and/or gross motor challenges, and delayed speech and language milestones.[3,6]

What are the common speech and language features in children with *FOXP2*-SLD?

A key feature of *FOXP2*-SLD is CAS (also known as developmental verbal dyspraxia or speech apraxia).[2, 3, 6] CAS is a motor disorder that affects the production, sequencing and stress of speech.[2,5] Many individuals with *FOXP2*-SLD have more than one speech disorder. For example, many children have CAS and a phonological disorder.[6]

Dysarthria, a neuromuscular speech disorder, has also been reported in a few individuals with a *FOXP2*-SLD, but this is less common than CAS.[3,6]

For some individuals, their speech challenges mean that they use augmentative and alternative communication (AAC) methods to support their communication. Some individuals use sign language, whilst others use graphic, aided AAC systems (e.g., communication books, high-tech speech generating devices).[6]

At what age do individuals with *FOXP2*-SLD begin speaking?

Many individuals have limited babbling as babies. More than half of individuals with *FOXP2*-SLD say their first words after 18 months old, which is older than is seen in typically developing children (12-15 months old).[6] Likewise, many individuals learn to combine words to create sentences later than seen in typical development (2-3 years), with many with *FOXP2*-SLD combining words as late as 8 years old, and some individuals not learning to combine words into adolescence.[6]

How can speech pathologists/therapists support children with *FOXP2*-SLD?

Currently, interventions are specific to an individual's communication needs. Individuals with *FOXP2*-SLD require a speech pathology or speech therapy assessment to ensure tailoring of best-evidenced interventions to the individual's profile.

Due to the high incidence of literacy challenges, alongside phonological difficulties, it is important that individuals with *FOXP2*-SLD receive systematic and intensive literacy supports to learn to write and read.[6]

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As speech and language disorders are a core feature of *FOXP2*-SLD, speech therapy/pathology input should start early in life and include **assessment** and **therapies** tailored to each individual. Many countries/states provide early intervention programs where speech therapy may be provided by government programs, educational programs, private practices, or a combination of these depending on your location. Families can seek advice from local practitioners about the services available to them in their region.

Assessment/evaluation

Important domains for a speech pathology assessment include:

- Speech production skills: to evaluate for specific speech diagnoses (e.g., CAS, phonological disorder)
- Expressive and receptive language skills
- Social/pragmatic language skills
- Feeding and swallowing abilities
- Literacy skills, such as phonological awareness, reading and writing

The types of assessment tools used will vary depending on the child's individual profile and developmental age. Assessment may be required at an initial diagnosis and throughout childhood and adolescence. The goal of assessment will be to understand the nature and severity of speech and language challenges, then make recommendations for appropriate therapies when needed.

Therapy/intervention

There is no research on speech and language interventions that are *specifically* designed for children with *FOXP2*-SLD. Speech and language interventions for children with *FOXP2*-SLD are currently guided by the child's individual profile and the best evidence for speech and language disorders more generally, and include:

• Augmentative and alternative communication (AAC)

AAC refers to ways of communicating other than talking (speech), such as the use of sign language or communication devices. AAC options can support language development prior to speech developing (using AAC does not prevent or slow down language development) and can also be of benefit when speech is unclear. Given children with *FOXP2*-SLD have delayed communication development, introducing AAC in the early years should be considered to foster language development and provide a means for children to engage, learn, and reduce communication frustrations. The need for AAC or the AAC options used by individuals may change over time. Speech pathologists/therapists work with children and families to find the most appropriate AAC options tailored to needs and abilities.

• Evidence-based treatments for CAS[7]

Existing treatments have varying levels of efficacy. Some examples include:

- Nuffield Dyspraxia Program[8]
- Rapid Syllable Transition Treatment (ReST)[8]
- Dynamic Temporal and Tactile Cueing (DTTC)[9]
- Prompts for Restructuring Oral Muscular Phonetic Targets (PROMPT)[10]

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Evidence based treatments for CAS are high-intensity, with at least weekly and sometimes multiple weekly speech therapy sessions.[11] See Morgan et al. (2018) Cochrane Review for more information on CAS therapies.[11] As, children with *FOXP2*-SLD are very likely to have CAS early speech therapy intervention is advised.

Families should ask their speech pathologist/therapist about how effective these programs (or the ones they are recommending) will be for their child given their age and symptoms. The type of therapy will depend on: (1) the child's symptoms, (2) their age, (3) the severity of their condition, and (4) any other health or development challenges they have.

Like with any skilled movement, practice or therapy is usually most successful when it happens several times a week. When CAS symptoms have resolved with therapy, there may still be a need for continued speech pathology/therapy input to address challenges in other areas of communication such as expressive language skills (e.g., vocabulary, sentence formation), social/pragmatic language skills (e.g., conversation skills, topic maintenance), and literacy.

It is also important to note that CAS is a difficulty with planning and programming movements for speech. There is no strong evidence to support the use of non-speech oral motor exercises alone (e.g., pursing, blowing, lip massage etc.) as an effective treatment for speech sound disorders.[12]

Do individuals attend mainstream school?

Most individuals with *FOXP2*-SLD attend mainstream schools with support including speech and language therapy. Some individuals attend special education schools.[4]

How does speech develop over time in *FOXP2-SLD*?

For adults with the *FOXP2*-SLD, whilst speech impairment is usually still evident, these challenges tend to lessen in severity as individuals grow older. Some speech sounds (e.g., the 'r' and 'th' sounds) and speech prosody remain challenging into adulthood for many individuals.[6]

We do not yet fully understand how speech develops over time for children with *FOXP2*-SLD, however studies are currently underway to learn more about the ongoing communication trajectory. To learn more about this study and get involved contact: angela.morgan@mcri.edu.au or speechtracker@mcri.edu.au.

Further information and support:

- More information on CAS: CAS Fact Sheet
- More information on phonological disorder: Phonological Disorder Fact Sheet
- More information on AAC: AAC Fact Sheet
- Unique *FOXP2*-SLD guide: *FOXP2*-SLD Guide
- Apraxia kids information support group: Support Group Website

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