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7q31 deletions affecting *FOXP2*
Fact sheet

What are 7q31 deletions?

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].

7q31 deletions can affect the *FOXP2* gene and neighbouring genes, which can result in altered development. Individuals with 7q31 deletions may also be referred to as having *FOXP2*+ related speech and language disorder.[2]

The size and location of an individual's 7q31 deletion will determine whether only speech and language is impacted (*FOXP2*-related speech and language disorder) or whether the individuals will experience broader (global) neurodevelopmental disabilities, such as intellectual disability and autism spectrum disorder (*FOXP2*-plus related speech and language disorder).[2,3]

Speech and Language

The terms 'speech' and 'language' are often used interchangeably; yet, they are categorised differently by a speech pathologist, which 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).

Childhood Apraxia of Speech (CAS) is a key feature in individuals with 7q31 deletions.[4] Approximately 1 in 3 children with CAS have a genetic cause for their CAS.[5] However, 7q31 deletions are very rare.[4]

The Information provided on this webpage pertains to 7q31deletions (as seen in *FOXP2*-plus related speech and language disorder) and not individuals with variants and deletions affecting only the *FOXP2* gene *(FOXP2*-related speech and language disorder).

What are the associated health and medical conditions seen in individuals with 7q31 deletions?[4]

- **Intellectual disability:** Individuals with large 7q31 deletions (>10Mb) are likely to have a moderate to severe intellectual disability. Individuals with smaller deletions (less than 10Mb), are more likely to have a mild intellectual disability. Some individuals with smaller deletions may not have an intellectual disability
- **Motor skill difficulties:** fine (e.g., writing) and gross (e.g., riding a bike) motor impairment
- **Delayed motor milestones:** individuals usually learn to walk after 16 months of age
- **Hypotonia** (low muscle tone)

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- **Feeding difficulties:** particularly in early childhood and increased difficulties with complex oral motor skills (e.g., licking food from lips)
- **Sleep disturbance:** such as difficulty falling asleep, frequent and early waking
- **Autism spectrum disorder**
- **Mental health diagnoses:** anxiety
- **Vision impairment**: namely long sightedness and strabismus

What are the common speech and language features in children with *FOXP2*-SLD?[4]

- **Speech disorder:** childhood apraxia of speech (CAS), a motor speech disorder causing disorders speech planning and programming is universal in all individuals who learn to speak.
- **Language disorder:** mild to severe language disorder across receptive (understanding) and expressive language skills. Those with smaller 7q31 deletions have stronger language skills than those with larger deletions.
- **Literacy difficulties:** challenges with reading and writing, ranging from severe impairment to average literacy skills.
- **Augmentative and alternative communication (AAC):** For some individuals, their speech and and language challenges mean that they use AAC to support their communication. Some individuals use key word sign, whilst others use aided AAC systems (e.g., communication books, speech generating devices).

At what age do individuals with 7q31 deletions begin speaking?

Individuals with smaller deletions often learn to speak after 18 months of age, later than what is seen in typical development (~12 months). Individuals with larger deletions (>12.5Mb) may not learn to speak, even into adulthood. Some individuals have learnt to speak first words, but may not learn to combine words into spoken sentences.[4]

How can speech pathologists/therapists support children, adolescent and adults with 7q31 deletions?

Currently, interventions are specific to an individual's communication needs. Individuals with 7q31 deletions require a speech pathology/therapy assessment to ensure tailoring of best-evidenced interventions to the individual's profile. As speech and language disorders are a core feature of 7q31 deletions, 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:

- Augmentative and alternative communication (AAC) e.g., key word sign, communication books, speech generative devices
- Speech production skills: to evaluate for specific speech diagnoses (e.g., CAS)
- Expressive and receptive language skills
- Social/pragmatic language skills
- Feeding and swallowing abilities

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- Literacy skills, such as systematic, synthetic phonics, and reading, and writing skills

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 individuals with 7q31 deletions. Speech and language interventions for for individuals with 7q31 deletions 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 individuals with 7q31 deletions have delayed communication development, and some may never learn to speak, 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[6,7]

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]

Evidence based treatments for CAS are high-intensity, with at least weekly and sometimes multiple weekly speech therapy sessions.[6,7] 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.

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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.[11]

Do individuals attend mainstream school?

Individuals with 7q31 deletions attend mainstream and specialist education settings with support including speech and language, occupational, and physiotherapy.[4]

How does speech develop over time in 7q31 deletions?

Some adults with 7q31 deletions, particularly those with larger deletions, will not learn to speak into adulthood, and will use AAC (e.g., key word sign) to communicate. For other adults with 7q31 deletions, they are likely to have persistent speech and language challenges into adulthood, but learnt to speak, write, and read.[4]

Further information and support:

- More information on CAS: CAS Fact Sheet
- More information on AAC: AAC Fact Sheet
- Apraxia kids information support group: Support Group Website

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