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Speech, language, and Koolen-de Vries syndrome

Miya St John and Prof Angela Morgan

What is Koolen-de Vries syndrome (KdVS)?

KdVS is caused by a change in the *KANSL1* gene. This is caused by either a small change within the *KANSL1* gene or a deletion on chromosome 17 (called a 17q21.31 deletion) which includes the *KANSL1* gene. A diagnosis of KdVS is established in an individual by genetic testing.

What are the associated health and medical conditions seen in KdVS?

Recent studies of KdVS indicate mild-severe intellectual disability (~85%), eye/vision changes (~60%), changes to the structure of the brain (~50%), dental problems (~50%), sleep disturbance (~40%), muscle/joint issues (~40%), changes to the structure of the heart (~40%), and epilepsy/seizures (35-75%). Other less common features include kidney/urogenital complications, hearing impairment, gastrointestinal problems.^{1,4}

What are the common speech and language features in children with KdVS?

Most individuals with KdVS communicate using verbal speech. Most children will rely on verbal speech by 6-7 years old.¹ However children do take some time to develop this.^{1,3} In KdVS, studies show that first words are often not achieved until 3 years old (sometimes much later).³ It is also reported that children show minimal or no babbling in the first 6 months of life.³

Studies have reported the following **speech disorders** in KdVS:

- 64-100% of individuals with KdVS who use verbal speech have **CAS**.^{1,3} CAS is most apparent during preschool and early school years.^{1,3}
- 46-93% of individuals with KdVS who use verbal speech have **dysarthria**.^{1,3} Dysarthria is most apparent during the later school years and adolescence.^{1,3}
- 77% of individuals with KdVS who use verbal speech **stutter**.¹ In KdVS, stuttering usually develops between 5-6 years of age (but can develop into adolescence). Stuttering in KdVS can come and go over time.^{1,3}
- Other speech disorders include **articulation** errors and **phonological** delay/disorder. These are usually milder.
- Many children with KdVS will experience a combination of the speech disorders above.

Speech vs Language

The terms 'speech' and 'language' are often used as the same terms; however, they actually mean different things to a speech pathologist:

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

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



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In terms of language, children with KdVS have difficulties organising and producing words and sentences (**expressive language impairment**) or understanding sentences and words (**receptive language impairment**).

Literacy (reading, spelling) and writing can be particularly challenging in KdVS.¹

Minimally verbal children with KdVS often communicate successfully using **alternative and augmentative communication (AAC)**. AAC refers to ways of communicating other than talking (speech), including sign language or communication devices. AAC can help individuals with KdVS to develop their language before their speech production ability develops.^{1,3}

Strengths in KdVS often include **positive social skills, behavioural and emotional control, and coping skills**.¹

Research shows that there is no difference in communication when comparing those with 17q21.31 deletions to those with *KANSL1* gene variants.¹

How can speech pathologists (SPs) support children with KdVS?

As speech and language disorders are a core feature of KdVS, SP 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 an SP 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

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 KdVS. Speech and language interventions for children with KdVS are currently guided by the child's individual profile and the best evidence for speech and language disorders more generally. Existing treatments have varying levels of efficacy. Some examples include:

CAS:

- Nuffield Dyspraxia Program⁵
- Rapid Syllable Transition Treatment (ReST)⁵
- Dynamic Temporal and Tactile Cueing (DTTC)⁶
- Prompts for Restructuring Oral Muscular Phonetic Targets (PROMPT)⁷



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Stuttering:

- Lidcombe programme⁸⁻¹⁰
- Demands and Capacities model⁸⁻¹⁰

Augmentative and alternative communication (AAC):

- 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 AAC options may change over time

Families should ask their SP 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 SP 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 of 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

How does speech develop over time in KdVS?

We do not yet fully understand how speech develops over time for children with KdVS, however studies are currently underway to learn more about the ongoing communication trajectory.

Further information and support:

- For information and support on KdVS: <https://kdvsfoundation.org>
- More information on CAS: [CAS Fact Sheet](#)
- More information on stuttering: [Stuttering Fact Sheet](#)
- More information on dysarthria: [Dysarthria Fact Sheet](#)
- More information on AAC: [AAC Fact Sheet](#)



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