



SPEECH, LANGUAGE, AND SETBP1-HD

What is SETBP1-HD?

SETBP1 is a gene located at chromosome 18q12.3.

SETBP1 haploinsufficiency disorder (SETBP1-HD) occurs when there is a change or small loss (microdeletion) within the *SETBP1* gene.

The diagnosis of SETBP1-HD is established in an individual by genetic testing.

What are the associated health or developmental conditions seen in children with SETBP1-HD?

SETBP1-HD is associated with motor developmental delay, speech and language disorder, attention-deficit/hyperactivity disorder (ADHD), and mild to moderate intellectual disability or learning difficulties.¹⁻⁴

What are the common speech and language features in children with SETBP1-HD?

The terms 'speech' and 'language' are often used interchangeably; however, they are separate entities:

Speech involves producing sounds in words with the correct breath support, voicing, resonance, articulation, prosody, and accuracy.

Language involves meaning, i.e., the understanding and expression of words (vocabulary) and sentences (grammar).

In terms of speech, children with SETBP1-HD take some time to develop their verbal speech. The most common speech disorder diagnoses in children with SETBP1-HD are typically childhood apraxia of speech (CAS) and phonological disorder.²

CAS is a difficulty with how the brain plans speech sounds. Children with CAS know what they want to say, but their brain struggles to correctly move their lips, jaw, and tongue to speak clearly and be understood.

Phonological disorder refers to difficulty understanding and using sounds correctly to convey meaning. Children with phonological disorder also often use sound error patterns that are typically seen in the speech of younger children (e.g., fronting of sounds such as 'tar' for car, reducing sound clusters such as 'bed' for bread etc.).

In terms of language, children with SETBP1-HD also have difficulties expressing themselves with regards to organising and producing words and sentences (expressive language impairment) or understanding sentences and words (receptive language impairment).

Individuals with this SETBP1-HD are sociable and have a strong desire to communicate.

Minimally verbal children with SETBP1-HD often communicate using sign language, gestures, or digital devices, which can help them to develop their language before their speech production ability develops.²

In what ways may speech-language pathologists (SLPs) support children with SETBP1-HD?

As speech and language disorders are a core feature of SETBP1-HD, SLP input should start early in life and include **assessment** of speech and language skills and **therapies** tailored to each child's individual profile and developmental age. 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 SLP assessment include:

- **Speech production skills:** to evaluate for specific speech diagnoses (e.g., CAS, phonological disorder)



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- Expressive and receptive language skills
- Social/pragmatic language skills

The types of assessment tools used may vary depending on the child's individual profile and developmental age, and assessment may be required both at initial diagnosis and at various stages throughout childhood and adolescence. The main goals of assessment will be to understand the nature and severity of each child's speech and language challenges to be able to make recommendations regarding appropriate therapies when warranted.

As well as working on speech and language, SLPs also assess and treat feeding and swallowing difficulties.

Therapies/interventions

There is no research on speech and language interventions that are *specifically* designed for children with SETBP1-HD. Speech and language interventions for children with SETBP1-HD 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 SETBP1-HD 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. SLPs work with children and families to find the most appropriate AAC options tailored to needs and abilities.

- **Evidence-based treatments for CAS**

Existing treatments have varying levels of efficacy for treating CAS, including:

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

Families should ask their SLP 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:

- the child's symptoms
- their age
- the severity of their condition, and
- any other health or development problems they have.

Like with any skilled movement, practice or therapy is usually most successful when it happens several times a week. While some children largely overcome CAS with therapy, others find therapy improves their ability to communicate but does not change the concentration CAS demands or the tiredness it can cause. For some children where CAS symptoms have been largely overcome with therapy, there may still be a need for continued SLP input to address challenges in other areas of communication such as expressive language skills (e.g. vocabulary, complex sentence formation etc.), social/pragmatic language skills (e.g. conversation skills, turn taking, topic maintenance etc.), and literacy.



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It is also important to note that CAS is a difficulty with planning 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.⁸

How does speech develop over time?

We do not yet fully understand how speech develops over time for children with SETBP1-HD, 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

- For information and support on SETBP1: <https://www.setbp1.org>
- More information on CAS: [CAS fact sheet](#)
- More information on phonological disorder: [Phonological disorder fact sheet](#)

References

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