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KIF1A-associated neurological disorder (KAND)

Fact sheet

What is KAND?

KIF1A-associated neurological disorder (KAND) is a genetic disorder. Common KAND characteristics include motor disorders, intellectual disability, visual impairment, brain changes and seizures.[1-4] KAND is caused by pathogenic or likely pathogenic variants in the *KIF1A* gene. The *KIF1A* gene codes for a protein called kinesin family member 1a. This protein is involved in transportation in the nervous system, including the brain.[5] There are many different types of *KIF1A* variants that can cause KAND. This means that individuals with KAND have a broad range of characteristics associated with the disorder.[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).

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

- Vision: many individuals have visual impairment, and some individuals may lose their vision over time due to optic nerve atrophy.[1-3]
- Motor disorders: some individuals have spasticity in their limbs, hypotonic core muscles, and difficulties coordinating movement. Many individuals seek support from occupational and physiotherapists. Individuals can lose motor skills and often require assistance to move around (e.g., a wheelchair).[1-3]
- Peripheral Neuropathy: individuals may experience reduced pain sensitivity, tingling in their extremities, and disrupted temperature regulation.[1,3]
- Brain changes: some individuals have brain changes called cerebellar and cerebral atrophy [1-3]
- Intellectual disability: most individuals have an intellectual disability, which can range from mild to profound.[1-4]
- Behavioural: some individuals also have attention deficit hyperactivity disorder (ADHD), autism and anxiety-related symptoms. [1,3]
- Regression: some individuals may be considered as having a childhood dementia, and experience brain deterioration.[1,3,4,6]
- Seizures: absence, tonic-clonic, and atonic seizures are the most common. Some individuals also experience epileptiform activity during sleep. Seizures are associated with worse cognitive function.[1-4]
- Bladder and Bowel Dysfunction.[1,3]: some individuals have urinary incontinence or gastrointestinal motility symptoms.

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At what age do individuals with KAND begin speaking?

Few individuals with KAND say their first words at a typical age (~12-months-old). Approximately one-quarter of individuals will say their first words after 18-months-old, and some individuals do not say their first words until they are a in primary/elementary school. [4]

Children also learn to combine words into sentences at a later age than expected. In typical development, children usually combine words into sentences by 2-years-old. However, most individuals with KAND will combine words into sentence after 4-years-old, and some individuals might never learn to combine words into spoken sentences. The location and type of *KIF1A* genetic variant may influence speech and language milestones.[4]

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

Approximately two-thirds of individuals with KAND will learn to combine words into spoken sentences. Of those individuals who learn to speak, many have speech disorders which impact their ability to be understood. The most common speech disorder is called dysarthria. Dysarthria is a neuromotor speech disorder. Individuals may present with dysarthria features such as slow speech rate, imprecise articulation, vocal harshness, hypernasality and decreased volume variation.[4]

Language skills vary greatly between individuals with KAND, although most individuals have some degree of receptive (understanding) and expressive (using language) impairment. Language abilities range from severely impaired to average. Overall, expressive language is a strength compared to receptive language. Written language skills are also generally low. Social communication can be challenging for individuals with KAND, although some individuals have average social communication skills. Most individuals with KAND are socially motivated, which is a significant strength. [4]

Individuals with few or no spoken word typically used conventional communication behaviours such as pushing/pulling and simple gestures (e.g., pointing) to communicate. These individuals were often able to refuse (say no), and make choices, but could rarely ask questions or make comments.[4]

How do speech and language features change over time in individuals with KAND?

Approximately 1/5th of parents report that their child with KAND has lost speech and language skills overtime.[4] Loss of adaptive behaviour skills and brain changes can also occur with age.[3]

Approximately half of individuals have used augmentative and alternative communication (AAC), such as key word sign or speech generating devices.[4] Individuals may use AAC throughout their life due to unclear speech or having few spoken words. Other individuals may just use AAC as a child, while their speech and language skills are developing.

How can speech pathologists/therapists support children with KAND?

There is no research on speech and language interventions that are specifically designed for individuals with KAND. At present, an individualised approach should be taken to assessment and management to ensure therapies are tailored to and optimised for each child.

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Due to delayed speech and language milestones in KAND, alongside dysarthria in individuals who use speech, AAC should be implemented as early as possible. AAC interventions often involve approaches such as Key Word Sign (e.g., Makaton, Baby Sign), communication books and boards, and speech generating devices. Speech generating devices (also known as voice output communication aides, electronic AAC or high-tech AAC) can enable the individual to communicate using an electronic voice when selecting icons or pictures on a digital screen. Speech generating devices can be on a dedicated electronic device, or be a specialised application on a general device, such as an iPad®. Additionally, some individuals use more than one AAC system or use an AAC system alongside speech. For some individuals, the disorder will progress. AAC can be helpful to support communication is there is a loss of speech and language skills. Consequently, AAC that is chosen should be adaptable to visual loss and motor changes, should they occur.

As many individuals have relative weaknesses in receptive language skills, strategies to support understanding such as using visual supports and repeating questions or instructions using simple language may be beneficial. Individuals with KAND will likely benefit from language intervention, including support with reading and writing where appropriate.

Do individuals attend mainstream school?

Some individuals attend mainstream school settings, whilst other individuals attend specialist school settings. A child's education setting is dependent on an individual child's support needs, alongside supports the education system around the child can offer.

Assessment/evaluation

Important domains for a speech pathology assessment include:

- Speech production skills: to evaluate for specific speech diagnoses (e.g., dysarthria)
- Expressive, receptive and pragmatic language skills
- Feeding and swallowing abilities
- Augmentative and alternative communication (AAC), e.g., communication aids
- Literacy assessment
- Assessment of appropriate environmental supports and practical communication needs of the child and their support people

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. Speech and language therapies should be tailored to an individual's communication support needs, be aware of regression associated with KAND.

Further information and support:

- For more information on speech and language research in KAND see our Plain Language Summary
- For more information on KAND: Health conditions caused by changes in the KIF1A gene Factsheet
- More information on dysarthria: Dysarthria Fact Sheet
- More information on AAC: AAC Fact Sheet
- KAND support groups: www.kif1a.org, www.kif1a.au

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