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CLN3 disease

Fact sheet

What is CLN3 disease?

CLN3 disease is a type of Batten disease. Batten disease is a group of genetic conditions which cause neurodegeneration.[1,2] CLN3 disease may be referred to as Juvenile Batten disease. Batten disease might also be known as Neuronal Ceroid Lipofuscinosis (NCL).

CLN3 disease is a type of childhood dementia which is autosomal recessive. Autosomal recessive means that individuals need two variants in both copies of the gene. In the case of CLN3 disease, individuals have a variant in each copy of the *CLN3* gene. The *CLN3* gene encodes for the CLN3 protein. The CLN3 protein helps cells digest and recycle different types of cell waste in the brain.

What are the associated health and medical conditions seen in CLN3 disease?

- Vision: individuals with CLN3 disease gradually lose their vision. This is usually the first sign of CLN3 disease.[1-3]
- Behavioural difficulties
- Motor disorders: many individuals seek support from occupational and physiotherapists. Individuals lose motor skills and require assistance to move around (e.g., a wheelchair).[3]
- Feeding difficulties: individuals often requiring enteral feeding (e.g., a G-tube) in the later disease stages (e.g., in their twenties).[3]
- Intellectual disability: individuals experience progressive cognitive decline.[1-3]
- Sleep disturbances[3]
- Epilepsy: individuals experience seizures which can become harder to control with medication.[1-3]

What are the initial speech and language features in children with CLN3 disease?

In the first few years of life, most individuals with CLN3 disease will show speech and language milestones like those seen in typical development. It is usually not until after the disease begins progressing in the

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).

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primary/elementary school years that speech and language disorders emerge. However, some individuals may exhibit speech and language disorders earlier in childhood.[3]

How do speech and language features change over time in children with CLN3 disease?

As the disease progresses, individuals experience changes in speech and language skills. Speech changes include speech becoming more difficult to understand due to a neuromuscular speech disorder called dysarthria. A common feature of dysarthria is speech dysfluency, also called neurogenic stuttering. Individuals may get stuck on the first sound, word or phrase in a sentence and repeat this part of the word/phrase. Language changes include losing grammar and vocabulary skills. Individuals may be able to say more (expressive language) than they can understand (receptive language).[3]

Individuals with CLN3 disease will usually end up losing all speech. Consequently, individuals may need an alternate way to communicate. Alternate ways to communicate are known as augmentative and alternative communication (AAC). AAC can include body movement, facial expression, on-body sign language, touch, and non-electronic (low-tech) and electronic (high-tech) communication aids.[3,4] By the time an individual reaches their twenties, they usually have very little verbal speech. Individuals usually pass away in their twenties.[1]

There are some individuals with CLN3 disease that might exhibit slower disease progression.[1,3]

How can speech pathologists/therapists support children with CLN3 disease?

There is no research on speech and language interventions that are specifically designed for individuals with CLN3 disease. At present, speech pathologists should take an individualised approach to both assessment and intervention, ensuring that therapies are tailored to and optimised for each child.

Due to the progressive nature of CLN3 disease, AAC should be implemented as early as possible alongside maintaining current speech and language skills for as long as possible. Due to the progressive loss of vision and motor skills, individuals may need to access communication aids via 'auditory partner-assisted scanning'. Auditory partner-assisted scanning refers to a communication partner reading the vocabulary options via the communication aid out loud, and the person with CLN3 disease indicating a 'yes' or a 'no' (e.g., via vocalising, gesture, body movement) if they want this vocabulary to be selected.[3,5]

To support families as communication changes overtime, caregivers and support people around the child should be provided with communication partner training.[6] Communication partner training may include strategies such as using simple language, breaking down instructions and tasks, using reminders and cues, providing choices, and using active listening strategies. Likewise, environmental supports should be considered to support an individual's understanding (receptive language), reduce disorientation, and support social connection with others.[7] Environmental supports should also consider visual impairment as a core feature of CLN3 disease. For instance, using auditory cues (e.g., songs and sounds) to communicate changes to routine, rather than visual cues.

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

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- Augmentative and alternative communication (AAC), e.g., communication aids
- Literacy assessment, e.g., Braille
- 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 and consider the progressive nature of the disease.[3,4]

Further information and support:

- For more information on speech and language research in CLN3 disease: Plain Language Summary
- More information on dysarthria: Dysarthria Fact Sheet
- More information on AAC: AAC Fact Sheet
- Batten disease support groups: https://bdsraaustralia.org/ https://bdsrafoundation.org/ http://www.bdfa-uk.org.uk/ https://www.battendisease.ca/

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