

Clinical picture of Kleefstra syndrome: speech, language and cognition in 103 individuals

Key terms

Genetics, neurodevelopmental, speech, language, intellectual disability, autism

What this research is about



Speech and language difficulties are common in Kleefstra syndrome. However, speech and language have not been researched in a group of people with Kleefstra syndrome. We wanted to understand the speech and language features, support needs and strengths in Kleefstra syndrome. This research helps us understand Kleefstra syndrome. This study

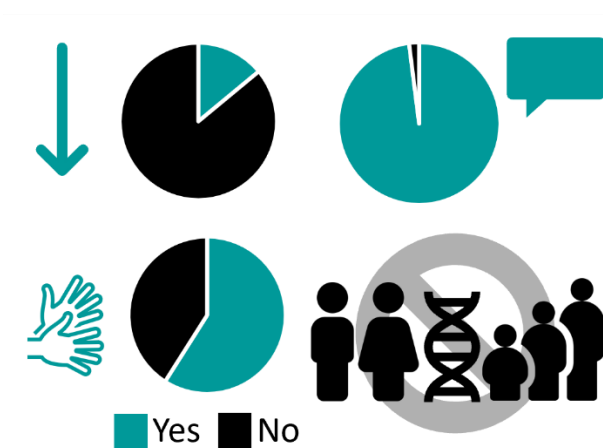
also helps us understand what the future holds for individuals, and what therapies and supports might be helpful individuals with Kleefstra syndrome.

What the researchers did



- 103 people with Kleefstra syndrome from 1 to 43 years old took part. They were from 26 countries.
- Health and development, feeding, adaptive behaviour, language, social communication, augmentative and alternative communication (AAC) and speech were assessed online.
- Speech was assessed via online video call for English-speaking participants.

What the researchers found



- 14 percent of people with Kleefstra syndrome have experienced regression, involving loss in language, motor and/or social skills.
- Language and cognition ranged from average to severely impaired in people with Kleefstra syndrome. 98 percent of verbal individuals had a speech disorder, particularly childhood apraxia of speech (CAS) and dysarthria.
- Augmentative and alternative communication (AAC, otherwise known as communication aids) was used by half the group, such as sign language and speech generating devices. AAC was important as many individuals had delayed language milestones, were minimally verbal, had hearing loss, had unclear speech, and/or had lost language skills due to regression.
- Genotype (apart from large >1Mb deletions), age, or sex did not cause differences in communication skills or adaptive behaviour.

What this means for people with Kleefstra syndrome and their families



- Regression can occur in Kleefstra syndrome, and it is important to look out for skill loss, particularly in adolescence and adulthood. Supports might have to adapt if regression occurs.
- Targeted speech therapy for childhood apraxia of speech (CAS) and dysarthria is important.
- Many people with Kleefstra syndrome would also benefit from early AAC access to support language development. AAC does not hinder natural speech development.

Learn more here: Morison LD, Kennis MG.P, Rots D, et al. J Med Genet 2024. doi:10.1136/jmg-2023-109702