# Kleefstra syndrome 1

Kleefstra syndrome 1, also known as KMT2C-related Kleefstra syndrome, is a rare genetic disorder that is primarily caused by mutations in the KMT2C gene. This syndrome is characterized by intellectual disability, developmental delays, and a range of other physical and neurological features. It is part of a group of related genetic syndromes known as “Kleefstra syndrome” or “Kleefstra spectrum disorders.”

Here are some key features and characteristics associated with Kleefstra syndrome 1:

Intellectual Disability: Individuals with Kleefstra syndrome 1 typically have moderate to severe intellectual disability, which affects their cognitive and adaptive functioning.

Developmental Delays: Children with this syndrome often experience delays in reaching developmental milestones, such as sitting up, crawling, walking, and speech.

Speech and Language Difficulties: Many individuals with Kleefstra syndrome have limited speech and language abilities, and some may be nonverbal.

Behavioral Issues: Behavioral challenges are common in Kleefstra syndrome, including hyperactivity, impulsivity, self-injurious behaviors, and social difficulties.

Physical Features: While there are no specific facial dysmorphic features associated with Kleefstra syndrome, individuals may have subtle facial differences, such as a long face or prominent chin.

Hypotonia: Low muscle tone (hypotonia) is often observed in affected individuals, which can contribute to motor delays.

Seizures: Some individuals with Kleefstra syndrome may experience seizures.

Recurrent Infections: There can be an increased susceptibility to respiratory infections.

Gastrointestinal Issues: Gastrointestinal problems, such as gastroesophageal reflux disease (GERD), constipation, and feeding difficulties, may occur.

Sleep Disturbances: Sleep-related issues, including difficulties falling asleep and maintaining regular sleep patterns, are common.

Growth and Feeding Problems: Some individuals may experience growth delays and feeding difficulties during infancy.

KMT2C gene mutations are typically de novo (occurring for the first time in the affected individual) and are not inherited from parents. These mutations can lead to changes in chromatin regulation and gene expression, affecting various aspects of development and function.

Management of Kleefstra syndrome 1 involves a multidisciplinary approach, including speech therapy, physical therapy, and behavioral interventions. Early intervention services and educational support are crucial to help individuals with this syndrome reach their full potential.

It's important to note that there is ongoing research in the field of genetics and rare genetic disorders, so additional information and advancements may continue to emerge regarding Kleefstra syndrome

1. Genetic counseling is recommended for families affected by this condition to understand the specific genetic changes and associated risks.

---

Niu M, Li Y, Zhan S, Sun B, Liu J, Wu Y. Tourette-like syndrome secondary to Kleefstra syndrome 1 with a de novo microdeletion in the EHMT1 gene. BMC Neurol. 2023 Oct 10;23(1):365. doi: 10.1186/s12883-023-03417-x. PMID: 37817104.