

## Communication in *CLCN4*-related neurodevelopmental condition (*CLCN4*-NDD)

### Key terms

*CLCN4*-NDD, speech, language, communication, augmentative and alternative communication (AAC)

### What is this research about?



Many people with *CLCN4*-NDD experience communication difficulties. They may experience challenges understanding and expressing language, which can make it hard to express needs and connect with others. However, speech and language has not yet been well described in people with *CLCN4*-NDD.

People with *CLCN4*-NDD often experience other difficulties, including learning challenges, motor (movement) impairments, and epilepsy. These factors can also affect how people with *CLCN4*-NDD learn and use language.

This study aimed to learn more about how people with *CLCN4*-NDD communicate, and the best ways to support their communication.

### What did the research involve?



- 13 people with *CLCN4*-NDD participated, from 6 different countries.
- Ages ranged from 1 to 41-years-old.
- Participants completed online surveys about health, development, cognition, language, and social communication.
- A speech pathologist assessed speech via an online video call.

## What did the researchers find?

- **All** participants had trouble understanding and expressing language, even as adults.
- Early communication milestones were **highly variable** in this group.
- 8 participants had **limited spoken language**, but used other ways to communicate (e.g., gesture, communication device).
- People with spoken language often had motor speech disorders including **Childhood Apraxia of Speech** and **dysarthria**.
- All participants were **intentional communicators**, being motivated to get their message across.
- There was a **big difference** between participants' ability to **express basic needs versus use communication for social interaction** and information seeking.



## What does this mean for people with *CLCN4*-NDD and their families?

- Speech and language assessment should be done early and focus on each person's **unique strengths and challenges**.
- AAC (such as picture boards or speech devices) should focus on helping people **express their needs, connect socially, and self-advocate**.
- Timely access to speech therapy focusing on **supporting motor speech skills** is recommended for people with *CLCN4*-NDD.



## Want to learn more?

Garrett, A., Kalscheuer, V. M., Figueroa, R. R., Palmer, E. E., & Morgan, A. T. (2025). *CLCN4*-Related Neurodevelopmental Condition: Characterization of Speech and Language Abilities. *American journal of medical genetics. Part A*, e64192. Advance online publication. <https://doi.org/10.1002/ajmg.a.64192>