# Speech Disorders in Individuals with 22q11.2 Duplication Syndrome – an Overview

There is less research information available on speech and voice disorders in 22q11.2 duplication syndrome (22q11.2DupS) than on the 22q11.2 deletion syndrome (22q11.2DS).

Palate abnormalities, and associated hypernasality (too much airflow through the nose while talking), occur at a much lower rate in the duplication than in the deletion.

Speech articulation disorders of all types are relatively common in the duplication as it is in the deletion, and voice disorders occur at a similar rate as the deletion.

## Common Disorders in 22q11.2DupS

- Voice
  - ❑ **Dysphonia** – Problem with voice production, often due to abnormalities of the vocal cord structure or function
- Speech sound disorders
  - ❑ Limited, impaired and delayed use of speech sounds
  - ❑ **Compensatory articulation disorder** – Atypical sound production using a different place of sound production
  - ❑ **Obligatory or passive errors** – Forced sound errors due to structural abnormalities in the speech system, such as VPD (example: /m/ for /p/ or /b/)
  - ❑ **Phonological disorder** – Difficulty learning the rules and patterns of speech sound production.
  - ❑ Developmental articulation disorders – Use of substitutions, omissions and distortions of the sounds of speech. Speech sound errors may persist longer than expected.
  - ❑ **Abnormal speech prosody** – Differences in speech rate, rhythm, and/or stress patterns
- Motor speech disorders
  - ❑ **Childhood apraxia of speech (CAS)** – Difficulty planning and coordinating movements needed for speech
  - ❑ **Dysarthria** – Weakness, paralysis, incoordination or abnormal tone in muscles of respiration, phonation, articulation and/or resonation
  - ❑ Mixed features of CAS and dysarthria
  - ❑ **Speech motor delay**

### Observations in small-scale research studies in 22q11.2DupS

- Almost all patients needed therapy due to delayed motor and speech-language milestones (Ref #1).
- The majority of patients who inherited the duplication from their parents (as opposed to having a new duplication) had speech-language delays (Ref #1).
- In one family where multiple members were affected by 22q11.2DupS, one child had speech delay as the only clinical finding for the syndrome (Ref #2).

### Recommended Evaluations

It is recommended that children with 22q11.2DupS be evaluated by a speech-language pathologist with follow up based on the child's needs.

Individuals should also have hearing evaluated routinely by an audiologist.

If there are delays or deficits, the child can have appropriate therapies as soon as possible.

## References / Resources


1. Cross-sectional and longitudinal findings in patients with proximal 22q11.2 duplication: A retrospective chart study – 2022
2. 22q11.2 duplications: Expanding the clinical presentation – 2022
3. Microduplication 22q11.2: a description of the clinical, developmental and behavioral characteristics during childhood – 2012
4. Clinical variability of the 22q11.2 duplication syndrome – 2008


22q International 22q11.2 Foundation

The mission of the International 22q11.2 Foundation is to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships.
This information is brought to you by the Foundation for educational purposes only. It is not intended to be taken as medical advice. If you have concerns, please talk to your healthcare provider.

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