Childhood motor speech disorders: who to prioritise for genetic testing

Key terms

Childhood Apraxia of Speech, dysarthria, speech motor disorder, genomic testing

What this research is about

1 in 1000 children have a motor speech disorder of childhood apraxia of speech (CAS) and/or dysarthria.

Recently, our research has shown ~ 1/3 children with motor speech disorder have a genetic explanation. Families also highly value genetic testing. Despite this, genetic testing is not routine in clinical care.

We wanted to further validate the diagnostic yield and find clinical variables that were predictive of a genetic diagnosis.

This research helps us to better prepare the clinical landscape for integration of genetic testing for childhood motor speech disorder.

What the researchers did

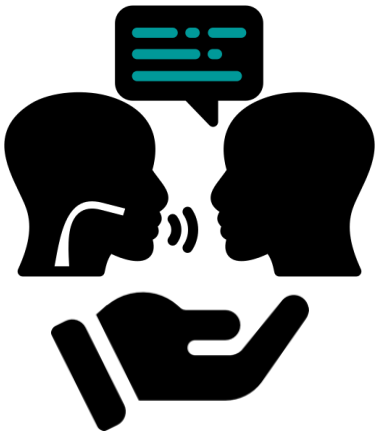
- 153 children took part in this study.
- Children were seen via in person appointment or online video call and assessed by a speech pathologist, clinical geneticist/paediatrician, and psychologist.
- Comprehensive genetic testing was performed once motor speech disorder was confirmed.

This summary was created following guidelines from Gasparini et al. (2025) *IJLCD.* https://doi.org/10.1111/1460-6984.13142

What the researchers found

- 44/153 **(29%)** children had a genetic diagnosis.
- This yield is comparable to disorders where genetic testing is **routine** (epilepsy, intellectual disability, cerebral palsy).
- Children that had **delayed walking, fine and gross motor disorder, receptive language impairment and/or cognitive impairment,** and **dysmorphism** were associated with having a genetic diagnosis.

What this means for people with motor speech disorder and their families



- Motor speech disorder has a comparable genetic diagnostic yield to other neurodevelopmental conditions, and genetic testing is important in finding an explanation.
- Genetic testing should be considered when a child presents with the identified clinical variables that are predictive of a genetic diagnosis.

Learn more here: Van Niel, H.*, Lauretta, M.*, Baker, E., O'Donnell, L., Boulton, C., Brenchley, C., Coman, D., Michellis, E., Goel, H., Thompson, G., Webster, R., Paxton, G., Stark, Z., Scheffer, I. E., Hildebrand, M. S., Amor, D. J., & Morgan, A. T. Childhood motor speech disorders: who to prioritise for genetic testing. *Eur J Hum Genet* (2026). https://doi.org/10.1038/s41431-025-01993-9
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