

MED13L-related disorder characterized by severe motor speech impairment

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Abstract

Background: MED13L-related disorder is associated with intellectual disability, motor delay, and speech deficits. Previous studies have focused on broad clinical descriptions of individuals, but limited information regarding specific speech diagnoses and results of direct testing has been published to date. We conducted deep phenotyping to characterize the speech, language, motor, cognitive, and adaptive phenotypes of individuals with MED13L-related disorder.

Methods: In this cross-sectional study, we administered standardized articulation, language, motor, and cognitive testing to 17 children and adolescents (mean age 9y 9m; SD 4y 5m; range 4y 2m to 19y 7m). In-person testing was supplemented with broad developmental, medical, and behavioral information collected virtually from a cohort of 67 individuals.

Results: All individuals who completed in-person articulation testing met diagnostic criteria for speech apraxia, dysarthria, or both. Language impairment was present in all of the in-person cohort and reported for almost all (97%) of the virtual cohort. Those who were able to complete motor testing demonstrated significant deficits in visual motor integration (mean 57.08, SD 9.26). Full scale IQs fell in the borderline to intellectual disability range, consistent with reported cognitive impairment in 97% of the virtual cohort. Notable medical features included hypotonia (83%), vision problems (72%), recurrent otitis media (58%), gastrointestinal problems (57%), and seizures (31%).

Conclusions: MED13L-related disorder is characterized by a high rate of motor speech disorders that occur in the context of globally impaired motor, language, and cognitive skills. Children would benefit from early referrals to speech therapy to assess their speech, language, and support needs.

Keywords: MED13L; Apraxia of speech; Dysarthria; Motor impairment; Speech disorders.

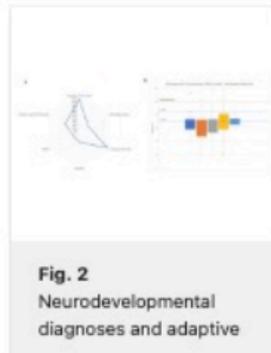
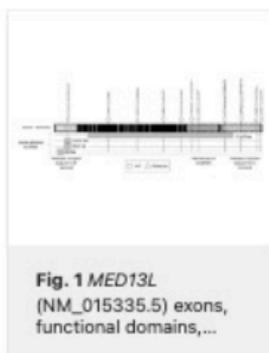
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Conflict of interest statement

Declarations. Ethics approval and consent to participate: Written informed consent for study participation was obtained from all participants or their legal representatives. Ethics approval for this study was obtained from the Geisinger Institutional Review Board (#00008345) under protocol #2013-0446. This research was conducted in accordance with the Declaration of Helsinki. Consent for publication: Not applicable. Competing interests: The authors declare no competing interests.

Figures



Update of

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