BRPF1-related disorder

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Pathogenic variants in BRPF1 cause intellectual disability, ptosis and facial dysmorphism. Speech and language deficits have been identified as a manifestation of BRPF1-related disorder but have not been systematically characterized. We provide a comprehensive delineation of speech and language abilities in BRPF1-related disorder and expand the phenotype. Speech and language, and health and medical history were assessed in 15 participants (male = 10, median age = 7 years 4 months) with 14 BRPF1 variants. Language disorders were common (11/12), and most had mild to moderate deficits across receptive, expressive, written, and social-pragmatic domains. Speech disorders were frequent (7/9), including phonological delay (6/9) and disorder (3/9), and childhood apraxia of speech (3/9). All those tested for cognitive abilities had a FSIQ ≥70 (4/4). Participants had vision impairment (13/15), fine (8/15) and gross motor delay (10/15) which often resolved in later childhood, infant feeding impairment (8/15), and infant hypotonia (9/15). We have implicated BRPF1-related disorder as causative for speech and language disorder, including childhood apraxia of speech. Adaptive behavior and cognition were strengths when compared to other monogenic neurodevelopmental chromatin-related disorders. The universal involvement of speech and language impairment is noteable, relative to the high degree of phenotypic variability in BRPF1-related disorder.