BRPF1-related disorder

https://pubmed.ncbi.nlm.nih.gov/37946714/

Bromodomain and PHD finger containing 1 (

BRPF1

)-related neurodevelopmental disorder is characterized by intellectual disability, developmental delay, hypotonia, dysmorphic facial features, ptosis, and blepharophimosis. Both

de novo

and inherited pathogenic variants have been previously reported in association with this disorder. We report two affected female siblings with a novel variant in

BRPF1

c.2420_2433del (p.Q807Lfs

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27) identified through whole-exome sequencing. Their history of mild intellectual disability, speech delay, attention deficient hyperactivity disorder (ADHD), and ptosis align with the features previously reported in the literature. The absence of the

BRPF1

variant in parental buccal samples provides evidence of a

de novo

frameshift pathogenic variant, most likely as a result of parental gonadal mosaicism, which has not been previously reported. The frameshift pathogenic variant reported here lends further support to haploinsufficiency as the underlying mechanism of disease. We review the literature, compare the clinical features seen in our patients with others reported, and explore the possibility of genotype-phenotype correlation based on the location of pathogenic variants in

BRPF1

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de novo

frameshift pathogenic variant in

BRPF1

in two siblings with this neurodevelopmental disorder.