Supplementary Table 2. Genetic variant details of patients with *KMT2B*-related dystonia

Patient code	Variant detail GRCh37	Exon	cDNA position NM_014727.3	Protein position	Zygosity	ACMG code	Pathogenicity	Novel
Patient 1	g.36228077A>G	Exon 33	c.7463A>G	p.Tyr2488Cys	Heterozygous	PM2PM6PP2-5	Likely pathogenic	No
Patient 2	g.36221499T>C	Exon 25	c.5258T>C	p.Leu1753Pro	Heterozygous	PM1PM2PP2-4	Likely pathogenic	No
Patient 3	g.36210725delC	Exon 3	c.481delC	p.Leu161fs	Heterozygous	PVS1PM2	Likely pathogenic	Yes
Patient 4	g.36214765delC	Exon 8	c.3192delC	p.Glu1065ArgTer117	Heterozygous	PVS1PM2PM6PP4	Pathogenic	Yes
Patient 5	g.36227669A>G	Exon 31	c.7238A>G	p.His2413Arg	Heterozygous	PM1PM2PP2PP4	Likely pathogenic	Yes
Patient 6	g.36223433G>A	Exon 28	c.5983G>A	p.Ala1995Thr	Heterozygous	PM2PP2PP4	VUS	Yes
Patient 7	g.36218074T>A	Exon 15	c.4021T>A	p.Cys1341Ser	Heterozygous	PM2PP2-4	Likely pathogenic	Yes

ACMG, American College of Medical Genetics and Genomics; PVS, very strong evidence of pathogenicity; PM, moderate evidence of pathogenicity; PP, supporting evidence of pathogenicity; VUS, variant of uncertain significance.