Table 1: Pathogenic variants in epilepsy associated genes.

Case	Gene	Type	Class	AA Change	cDNA Variant †	CADD Phred	Polyphen-2 Scores ‡	Epilepsy	Infantile Spasms
1	***CASK***	Inherited (mat)	Missense	R618K	c.1853G>A	21.4	1.0 / 1.0	Yes	No
2	***MAGI2***	*De novo*	Splice	-	c.3795+3T>G	21.1	-	Yes	Yes
3	***PRRT2***	Inherited (mat)	Missense	P362T	c.1084C>A	24.4	0.99 / 0.94	Yes	Yes
4	***RBFOX3***	*De novo*	Missense	A207V	c.620C>T	34.0	0.98 / 0.67	Yes	Yes
5	***CACNA1H***	Inherited (mat)	Missense	P618L	c.1853C>T	25.0	0.89 / 0.53	Yes	No
	KCNT1	Inherited (mat)	Missense	V296M	c.886G>A	27.6	1.0 / 1.0		
6	***RYR3***	Inherited (pat)	Missense	G592R	c.1774G>A	32.0	1.0 / 1.0	Yes	Yes
7	***CPA6***	Inherited (mat)	Nonsense	R311X	c.931C>T	46.0	-	No	-
8	***POLG***	Inherited (pat)	Missense	M1116V	c.3346A>G	26.1	0.98 / 0.98	No	-

† HGVSc notation (mRNA transcript reference in supplemental table 1.

‡ Polphen-2 scores presented as HumDiv / HumVar.

Abbreviations: AA = amino acid; mat = maternal; pat = paternal.