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Supplemental Data

A Homozygous Mutation in Human *PRICKLE1* Causes an Autosomal-Recessive Progressive Myoclonus Epilepsy-Ataxia Syndrome

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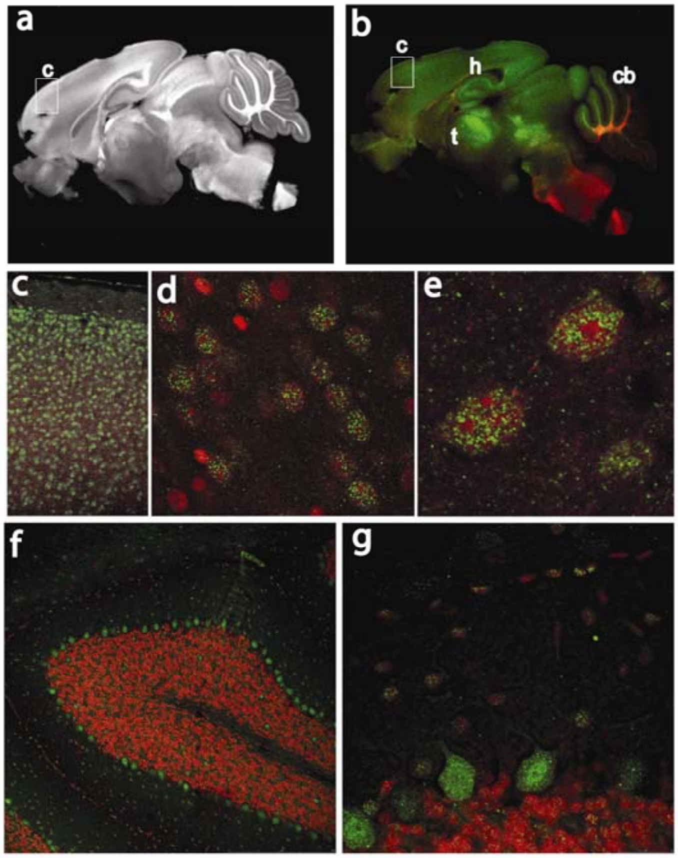

Figure S1. Prickle1 expression in P19 mouse brain. Light microscopy (a) shows saggital block used for these sections. Specific brain regions are marked (b) including c=cortex, h=hippocampus, t=thalamus, and cb=cerebellum. Low power (c) and increasingly higher power sections (d,e) from cortex, and low power (f) and higher power sections from cerebellum. Prickle1 is in the green channel and DNA stain is in red.

Table S1. Candidate genes sequenced prior to discovering *PRICKLE1* as the causative gene for progressive myoclonic epilepsy with ataxia.

Gene	Name	Gene ID	OMIM
ASB8	Ankyrin repeat and SOCS box-containing 8	140461	
ALG10	Asparagine-linked glycosylation 10 homolog	84920	603313
ALG10B	Asparagine-linked glycosylation 10 homolog B	144245	
ABCD2	ATP-binding cassette, sub-family D, member 2	225	601081
NELL2	Nel-like 2	4753	602320
SLC38A2	Solute carrier family 38, member 2	54407	605180
LRRK2	Leucine-rich repeat kinase 2	120892	609007
SLC2A13	Solute carrier family 2, member 13	114134	611036
FLJ20489	Hypothetical protein	55652	
GLT8D3	Glycosyltransferase 8 domain-containing 3	283464	
CNTN1	Contactin 1	1272	600016
SLC38A1	Solute carrier family 38, member 1	81539	608490
SLC38A4	Solute carrier family 38, member 4	55089	608065
DBX2	Developing brain homeobox 2	440097	
ADAMTS20	ADAM metallopeptidase with thrombospondin type 1 motif, 20	80070	611681
AMIGO2	adhesion molecule with Ig-like domain 2	347902	
ARID2	AT rich interactive domain 2 (ARID, RFX-like)	196528	609539
BICD1	bicaudal D homolog 1 (Drosophila)	636	602204
C12ORF40	chromosome 12 open reading frame 40	283461	
CPNE8	copine VIII	144402	
DNM1L	dynamin 1-like	10059	603850
FAM113B	family with sequence similarity 113, member B	91523	
FGD4	FYVE, RhoGEF and PH domain containing 4	121512	611104
RPAP3	RNA polymerase II associated protein	79657	611477
HDAC7A	histone deacetylase 7A	51564	606542
IRAK4	interleukin-1 receptor-associated kinase 4	51135	606883

KIF21A	kinesin family member 21A	55605	608283
MUC19	mucin 19, oligomeric	283463	612170
P11	26 serine protease	8909	606720
PDZRN4	PDZ domain containing RING finger 4	29951	609730
PKP2	plakophilin 2	5318	602861
PLEKHA9	pleckstrin homology domain containing, family A member 9	51054	
PPHLN1	periphilin 1	51535	608150
TWF1	twinfilin, actin-binding protein	5756	610932
PUS7L	pseudouridylate synthase 7 homolog (S. cerevisiae)-like	83448	
RAPGEF3	Rap guanine nucleotide exchange factor (GEF) 3	10411	606057
SFRS2IP	splicing factor, arginine/serine-rich 2, interacting protein	9169	603668
SYT10	synaptotagmin X	341359	
TMEM106C	transmembrane protein 106C	79022	
TMEM117	transmembrane protein 117	84216	
TMEM16F	transmembrane protein 16F	196527	608663
VDR	vitamin D receptor	7421	601769
YAF2	YY1 associated factor 2	10138	607534
YARS2	tyrosyl-tRNA synthetase 2 (mitochondrial)	51067	610957
ZCRB1	zinc finger CCHC-type and RNA binding motif 1	85437	610750

Table S2. Primers and conditions for *PRICKLE1* sequencing. F= forward primer. R=reverse primer. NF=nested forward primer. bp=basepairs.

Primer sequence		Product Size	Annealing Temperature
EXON1	F-GCC GGA AGG AAT CTA AGT GT	1040bp	52˚C
	NF-TTT TCT CCC TGC GCA TAA TC		
	R-CAG CCC AAC AAA CCG ATT AT		
EXON2	F-GGT CGG GGG TAA GAG AAA TG	353bp	60.5˚C
	R-TGG TAT TCC AGC ATC TCA GTG		
EXON3	F-AGA GGC CAA ACC CTG TAC CT	303bp	60.5˚C
	R-GGA GTT GGG GTT TAT GAG CA		
EXON4	F-TTC CCT TTT TCT AGA GAG GCT GT	400bp	60.5˚C
	R-TGC TAG TCC AGT CAC CTA CCC		
EXON5	F-AGG AAA GCC TGA GAA TCC TG	350bp	60.5˚C
	R-ATT TTG CTT GAT GTA AAC AGT GGA		
EXON6	F-TTT AAG AGC CAG TGT CTG TCC A	452bp	62˚C
	NF-CCC GGA AAA CAT GCA GTT AC		
	R-CAA AGC TCA TCA GCT GGA AC		
	NR-CTA GGT GAC AGG GCA GAC G		
EXON7A	F-GCT CCC CCA TAC CCA TAA TC	451bp	60.5˚C
	R-TTC GAG AAAGGG TGT CAT CA		
EXON7B	F-CAA GTT TCC TGG CCT CTC AG	367bp	60.5˚C
	R-CAG TCC ATC TTG TGA CTG TGC		
EXON7C	F-CCA GAG CCT TGC AAG TAA AAA	350bp	60.5˚C
	R-ACT GCG CCT GGC TTG AAT		
EXON8A	F-TTG AGA TTG GAA ATT TTC TTT GAA	416bp	60.5˚C
	R-TGC CGG ATT TCA ATG TCA TA		
EXON8B	F-ACC CCA GCA GGT CAA GTT TT	424bp	64˚C
	R-GGA GGA GGA GGA AGA ACA CC		
EXON8C	F-TGA ATC GGT TTCTGG GAC TC	369bp	62˚C
	R-ACA TGG GCA AAG AAA GCA CT		