The American Journal of Human Genetics, Volume *101*

Supplemental Data

Mutations in *TRAPPC12* Manifest in Progressive Childhood Encephalopathy and Golgi Dysfunction

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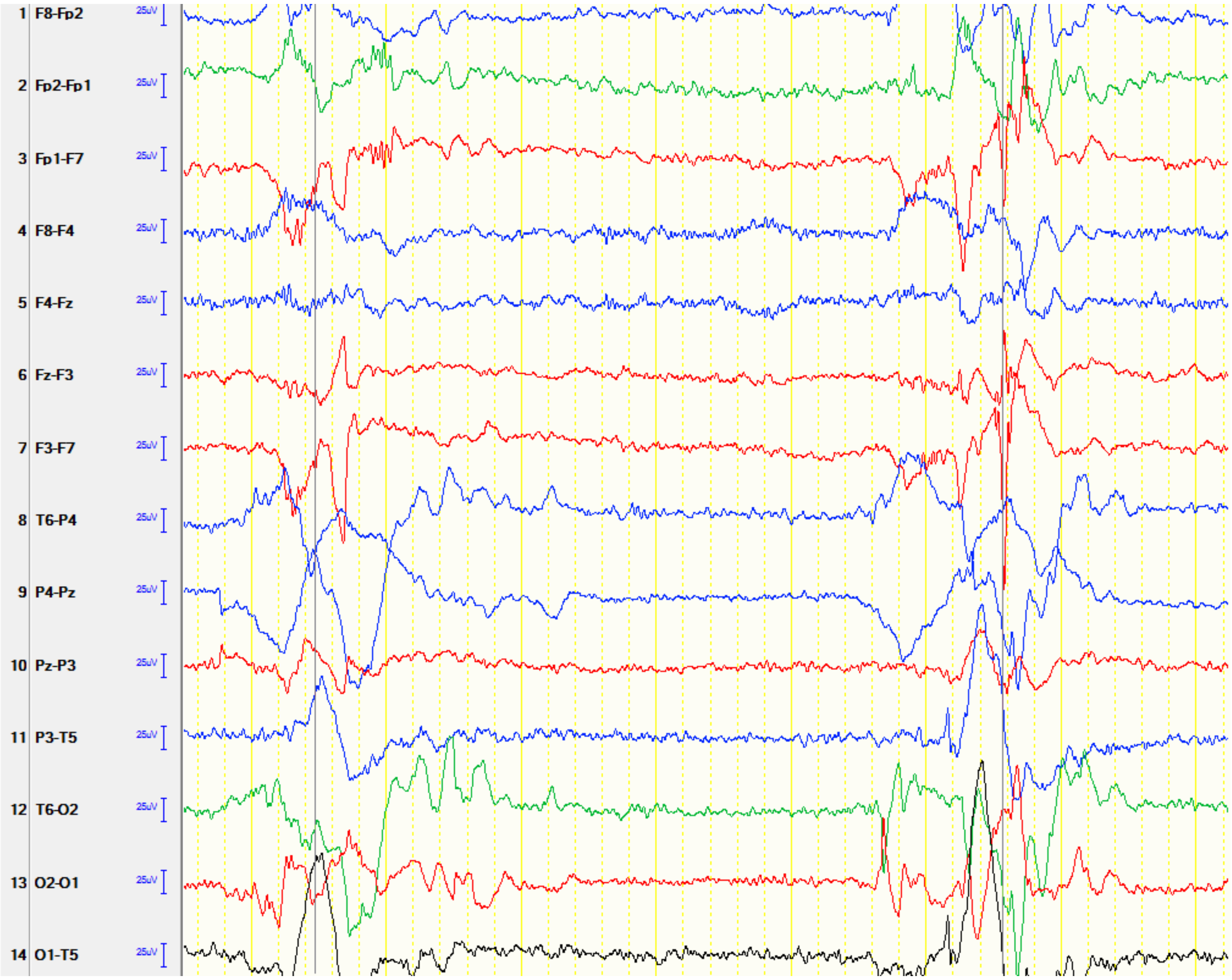

Figure S1. EEG at six months of age in 1:II-8 shows disorganized background activity with multifocal and generalized epileptiform activity consistent with hypsarrhythmia.

Table S1. Candidate recessive genetic causes based on exome sequencing in families F4331 and UW336

F4331 Family

Chr	bp (hg19)	ref	alt	gene	cDNA change	AA change	ExAC MAF	GERP	CADD	MT	rs
2	3391538	CG	C	*TRAPPC12*	NM_016030.5:c.145del	p.Glu49Argfs*14	NR	5.00	16.18	D	NR
3	126070876	C	A	*KLF15*	NM_014079.3:c.890G>T	p.Gly297Val	NR	4.25	10.94	D	NR
3	126722227	G	A	*PLXNA1*	NM_032242.3:c.1432G>A	p.Val478Ile	0.0008	4.43	17.75	D	rs199693063
3	148601571	C	T	*CPA3*	NM_001870.2:c.950C>T	p.Thr317Ile	0.0018	4.77	20.2	D	rs142588358
5	56155651	G	A	*MAP3K1*	NM_005921.1:c.743G>A	p.Arg248Gln	0.0002	5.72	20.5	D	rs201579608
7	111926991	T	C	*ZNF277*	NM_021994.2:c.155T>C	p.Leu52Ser	0.000008	1.97	13.74	P	rs764056350
8	10467637	T	C	*RP1L1*	NM_178857.5:c.3971A>G	p.Glu1324Gly	NR	-1.85	6.54	P	rs4240659
12	49725054	T	C	*TROAP*	NM_005480.3:c.2156T>C	p.Leu719Ser	0.0001	4.73	17.59	P	rs545155075
13	52676340	T	C	*NEK5*	NM_199289.1:c.698A>G	p.His233Arg	0.004	-7.44	0.009	P	rs56369842
13	52710344	C	T	*NEK3*	NM_001146099.1:c.1031G>A	p.Arg344Gln	0.0016	0.85	8.65	P	rs74087069
X	48460322	G	A	*WDR13*	NM_017883.4:c.982G>A	p.Val328Ile	0.0005	3.65	0.18	P	rs150193416
X	63409994	G	A	*AMER1*	NM_152424.3:c.3173C>T	p.Pro1058Leu	0.00002	4.83	17.58	D	rs760083711
X	73641702	C	A	*SLC16A2*	NM_006517.4:c.230C>A	p.Pro77His	NR	2.24	12.49	P	NR
X	101139020	T	G	*ZMAT1*	NM_001011657.3:c.1379A>C	p.Gln460Pro	NR	3.22	13.25	P	NR
X	117758578	G	C	*DOCK11*	NM_144658.3:c.3548G>C	p.Gly1183Ala	0.00001	5.53	6.79	D	rs370985549

UW336 Family

Chr	bp (hg19)	ref	alt	gene	cDNA change	AA change	ExAC MAF	GERP	CADD	MT	rs
2	3391754	C	CC	*TRAPPC12*	NM_016030.5:c.360_361insC	p.Glu121Argfs*7	NR	1.09	9.89	D	NR
2	3482619	C	T	*TRAPPC12*	NM_016030.5:c.1880C>T	p.Ala627Val	0.000008	5.92	21.3	D	rs768950892
21	37581084	G	A	*DOPEY2*	NM_005128.2:c.563G>A	p.Ser188Asn	0.008263	4.11	20.2	P	rs142091518
21	37597909	C	G	*DOPEY2*	NM_005128.2:c.1417C>G	p.Pro473Ala	0.008141	5.05	17.66	P	rs138343054

alt=alternate base, bp=base pair position (hg19 reference genome), CADD=Combined Annotation Dependent Depletion version 1.0, Chr=Chromosome, D=Disease Causing per Mutation Taster, ExAC MAF=Exome Aggregation Consortium Minor Allele Frequency, GERP=Genomic Evolutionary Rate Profiling, NR=Not Reported, P=Polymorphism per Mutation Taster, ref=reference base, rs= Reference SNP cluster ID