Supplemental Table 3: Homozygous variants in P1 and P2 identified by exome sequencing, Related to Figure 1 and the Human Subjects section of the STAR Methods.

Gene	Genom. Pos (hg19)	Protein change	CADD-score	gnomAD pLI	gnomAD MAF
EMP1	12:13364476T>G	V11G	26.7	0.5	0.001132
N6AMT2	13:21331602C>T	G45K	26.9	0	NA
PDE3A	12:20523148G>T	R310S	26.8	0	0.00001284
CCR8	3:39374669C>T	H283Y	23	0.01	NA
TBK1	12:64890825G>A	W619*	39	1	NA

Genom. Pos: genomic coordinates of variant in the human reference assembly (GRCh37/hg19); CADD score, Combined Annotation Dependent Depletion deleteriousness score; gnomAD pLI, Genome Aggregation Database probability of being loss-of-function intolerant score; Genome Aggregation Database minor allele frequency