Supplementary Material

We extracted genomic DNA from peripheral-blood leukocytes in four individuals (II:1, II:2, III:2, and III:3) and subjected it to whole-exome capture and sequencing on the Agilent SureSelect Human All Exon V6 (Agilent Technologies, Santa Clara, CA, USA) and the Illumina HiSeq2500 sequencing platform, respectively. Raw sequence reads were processed and aligned to the hg19 human reference sequence with the Burrows-Wheeler Aligner (version 0.7.12). Duplicate reads were removed with Picard, and local alignment optimization was performed with the Genome Analysis Tool Kit (GATK, version 3.4.0). The single nucleotide polymorphism and short indel candidates were annotated by SnpEff (version 4.1g). After screening of epilepsy-related genes, we identified a novel deletion variant (NM_001184880.1:c.592_599del) in *PCDH19*, which was predicted to result in a frameshift and premature termination of the PCDH1 protein (p.Arg198Alafs*25). We subsequently validated this variant by Sanger sequencing. This variant was not found in the Korean Reference Genome Database or the >138,000 exome and genome sequencing data deposited in the Genome Aggregation Database.