

eFigure 1: The c.2802-1G>C variant induces aberrant DEPDC5 splicing. (A) RT-PCR analysis of the *DEPDC5* spliced transcripts on a 1% agarose gel. RT-PCR products were amplified from total mRNA derived from blood and fresh brain. Shown in the white rectangle are the abnormal 1700-2000-bp fragments in individuals carrying the c.2802-1G>C variant (patient 4 and her mother) consistent with intron 29 retention, and the normal 500-bp fragment. (B) Sashimi plot shows RNA-seq mapping to *DEPDC5* and intron 29 retention in patient 4 and her mother.

