



**Supplementary Figure 1.** Genomic structure and mRNA expression of the human *SHANK3* gene. **(a)** Genomic structure of the *SHANK3* gene, which is located on chromosome 22q13.3, spans 57 kb and contains 24 exons. The mouse gene on chromosome 15qE3, the rat gene on chromosome 7q34, and the human gene share the same genomic structure. **(b)** Specific RT-PCRs were performed on total RNA from different brain regions and cDNA libraries (Clontech) to detect *SHANK3* mRNA and the alternative splice of exon 18, exon 22a, b and c. The age of the male and the two females studied were 74, 55, and 36 years, with a post-mortem delay of 10, 24 and 2 h, respectively. FC, frontal cortex; TC, temporal cortex; OC, occipital cortex; H, hippocampus; T, thalamus; C, cerebellum. Normal control human brains were obtained at autopsy under guidelines approved by the ethics committee. BLCL: B lymphoblastoid cell lines.