



**Fig. S1. Sequence of SASH1 gene.** (a) The proband's grandfather with normal sequence from exon 13 of SASH1 gene. (b) The proband's grandmother with normal sequence from exon 13 of SASH1 gene. (c) The proband with a heterozygous transition mutation c.1519T>G, resulting in missense mutation p.Ser507Ala. (d) His affected mother with the same mutation. (e) Part of the normal sequence from exon 13 of the SASH1 gene. Arrows indicate site of mutation (c.1519T>G).