



*Case Report*

## Mutation screening of *TSC1* and *TSC2* genes in Chinese Han children with tuberous sclerosis complex

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**ABSTRACT.** Tuberous sclerosis complex (TSC) is an autosomal dominant neurogenetic disorder caused by mutations in the *TSC1* or *TSC2* genes and is frequently associated with hamartoma formation in multiple organ systems. Here, we report two novel mutations in the *TSC2* gene, including a splicing mutation (IVS 29 +1G>C) in intron 29 and a deletion/insertion mutation (C.5090-5092delCCA- inAG) in exon 39 in two Chinese Han children with TSC whose first clinical manifestation was seizure. The identification of these two mutations confirmed the diagnosis of TSC and expands the spectrum of *TSC2* mutations causing TSC.

**Key words:** *TSC1* gene; *TSC2* gene; Tuberous sclerosis complex; Mutation