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Case Report

A novel *TSC1* mutation (c.1964delA) in a Chinese patient with tuberous sclerosis complex

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ABSTRACT. Tuberous sclerosis complex is an autosomal-dominant heritable disease caused by mutations in the *TSC1* and *TSC2* genes. We studied a Chinese patient with sporadic tuberous sclerosis complex. The clinical features of this patient included epilepsy, hypomelanotic macules and angiofibromas on his back; a cranial CT scan showed subependymal nodules along the lateral walls of the lateral ventricles. The *TSC1* and *TSC2* genes were studied by PCR and direct sequencing of the entire coding region and exon-intron boundaries of these genes. A novel deletion mutation (c.1964delA) in the *TSC1* gene exon 15 was identified, which was not present in his parents or 100 unrelated normal controls. This is the first report of this c.1964delA mutation of the *TSC1* gene, associated with tuberous sclerosis complex, expanding the spectrum of *TSC1* mutations that cause this disease.

Key words: Tuberous sclerosis complex; Mutation; *TSC1*; Gene

