Case Report

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

G.-X. Wang¹,², D.-W. Wang¹, J.-S. Zhao³, S.-F. Wang² and R.-P. Sun¹

¹Department of Paediatrics, Qilu Hospital of Shandong University, Jinan, P.R. China
²Institute of Paediatrics, Qilu Children’s Hospital of Shandong University, Jinan, P.R. China
³School of Life Science, Shandong Normal University, Jinan, P.R. China

Corresponding author: R.-P. Sun
E-mail: gxw5201@163.com

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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