GENETICS AND GENODERMATOSES

MUTATION ANALYSIS OF TSC1 AND TSC2 GENE IN PATIENTS WITH TUBEROUS SCLEROSIS COMPLEX

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Background: Tuberous sclerosis complex(TSC) is an autosomal dominant genetic disease. Which was caused by mutations in TSC1 or TSC2 gene. If mutations happened in either TSC1 or TSC2 gene, it would result in the occurrence of TSC. In the study, we collected the clinical data from three families and two sporadic cases of TSC of Han nationality in Sichuan Province, China.

Objective: To identify pathogenic mutations of TSC1 and TSC2 gene in three families and two sporadic cases with tuberous sclerosis complex (TSC) of Han nationality in Sichuan Province, China.

Materials and Methods: We collected blood samples and clinical data from three families and two sporadic cases. Genomic DNA was extracted from peripheral blood of the proband and the sporadic cases. Polymerase chain reaction (PCR) was used to amplify all the exons and exon-intron flanking sequences of TSC1 and TSC2 genes, and the products were analyzed by direct sequencing. Finally the results were analysed by Chromas 2.0 to identify new pathogenic mutations.

Results: One novel missense mutation c.1964C>T(P.S655F) from the first familial case was detected in the exon 19 of the TSC2 gene; One novel mutation c.1119-1120insA (p.S374KfsX2) from the first sporadic case was detected in the exon 11 of the TSC1 gene; One repeat deletion mutation c.5238-5255delCATCAAGCGGCTCCGCCA (p.his1746GlnfsX56) from the second sporadic case was detected in the exon 40 of the TSC2 gene.

Conclusion: The novel missense mutation c.1964 C>T(P.S655F) and the novel insertion mutation c.1119-1120insA(p.S374KfsX2) may be the underlying cause of the first familial and the first sporadic case with TSC respectively. The deletion mutation 5238-5255delCATCAAGCGGCTCCG CCA (p.his1746GlnfsX56) of TSC2 may be the underlying cause of the second sporadic case with TSC.