

Tuberous Sclerosis Complex Confirmed by Genetic Analysis: A Case Report

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Introduction

Tuberous sclerosis complex (TSC), also known as Bourneville disease, is an autosomal-dominant, neurocutaneous, multisystem disorder [1]. The underlying genetic cause is mutations in the *TSC1* or *TSC2* gene, which leads to overactivation of the mammalian target of rapamycin (mTOR) protein complex [2,3]. The main clinical features of the disease are facial sebaceous adenomas, epilepsy, and hypophrenia. Currently diagnosis is based on the criteria established at the 2012 International Tuberous Sclerosis Complex Consensus Conference [4]. mTOR inhibition is presently the main treatment [2,5]. It is still difficult to differentially diagnose TSC from other conditions such as neurofibromatosis, epilepsy, and mental disease, which makes genetic analysis important in its diagnosis.

Case Report

We report the case of patient with TSC associated with a novel mutation in the *TSC2* gene. In May 2015, a 31-year-old man was admitted to our hospital with a 7-months history of sudden-onset loss of consciousness, characterized by lack of responsiveness to any external stimuli, and sometimes accompanied by urinary incontinence. Each attack lasted for 4-5 seconds, with a frequency of 1-2 episodes a day. The patient was unaware of the attack, and often felt dizziness before onset. He visited our outpatient clinic, and was diagnosed with secondary epilepsy. After treated with oxcarbazepine for a week, he did not experience any further attacks. His previous medical history, personal history, and family history revealed no significant findings. Physical examination revealed many sebaceous adenomas on the face. Results of a neurological examination were normal.

MRI showed multiple patchy signal abnormalities in the brain, and round abnormal signal intensity deep in the left temporal lobe (**Figure 1**). CT revealed similar abnormalities (**Figure 2**). Significant abnormal recordings were observed on long-term video EEG. Abdominal Doppler ultrasound showed multiple hyperechoic tubers in the liver. Results of a CSF examination were

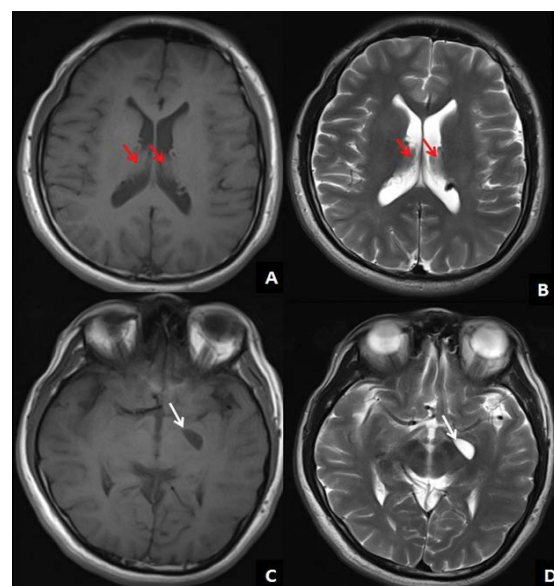


Figure 1 Axial MRI of the skull. Axial MRI showing (A and B) multiple patchy signal abnormalities in the brain (red arrows), (C and D) oval abnormal signal intensity deep in the left temporal lobe (white arrows).

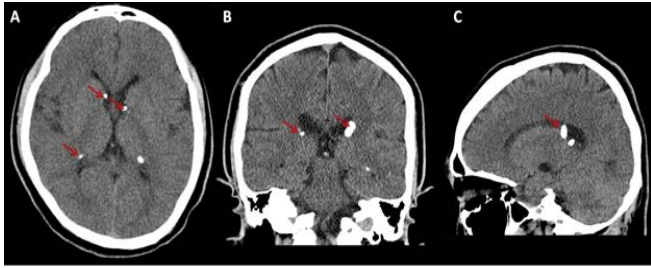


Figure 2 CT images. Axial (A), coronal (B), and sagittal (C) CT image showing multiple patchy signal abnormalities in the brain (red arrows).

normal with the exception of mild elevation of IgA, IgM, and IgG. Genetic analysis of a blood sample revealed a novel mutation in the *TSC2* gene (*TSC2* c.3355C>T p. (Gln1119*) heterozygosis). Based on these findings, the patient was diagnosed with TSC.

Discussion

According to the diagnostic criteria mentioned previously, this case displayed two major features and one minor feature. Therefore, it can be classified as a “definite diagnosis,” which was confirmed by genetic testing. Although this patient visited our hospital for epilepsy treatment, we detected a novel mutation in the *TSC2* gene. Therefore, it is necessary for doctors to be familiar with the clinical features of TSC to confirm the diagnosis with genetic testing if possible.

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References

- 1 DiMario FJ, Sahin M, Ebrahimi-Fakhari D (2015) Tuberous Sclerosis Complex. *Pediatr Clin North Am* 62: 633-648.
- 2 Curatolo P (2015) Mechanistic target of rapamycin (mTOR) in tuberous sclerosis complex-associated epilepsy. *Pediatr Neurol* 52: 281-289.
- 3 Wataya-Kaneda M (2015) Mammalian target of rapamycin and tuberous sclerosis complex. *J Dermatol Sci* 79: 93-100.
- 4 Northrup H, Krueger DA (2013) Tuberous sclerosis complex diagnostic criteria update: recommendations of the 2012 International Tuberous Sclerosis Complex Consensus Conference. *Pediatr Neurol* 49: 243-54.
- 5 Tran LH, Zupanc ML (2015) Long-Term Everolimus Treatment in Individuals with Tuberous Sclerosis Complex: A Review of the Current Literature. *Pediatr Neurol* 53: 23-30.