英文題目:Successful Control with Stiripentol of One Patient with Dravet Syndrome

中文題目:以 stiripentol 成功地控制 Dravet syndrome: 一病例報告

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Abstract

Background: Dravet syndrome (DS) is an early-onset epileptic encephalopathy characterized by generalized tonic, clonic, and tonic-clonic seizures that are initially induced by fever and begin during the first year of life. Seizures are usually refractory to treatment. Mutations of the *SCN1A* gene were found in a high percentage (range 35-100%) of patients with DS. Recently, the efficacy of stiripentol is proven in Dravet syndrome. We reported a patient who was diagnosed with DS, based on electroclinical symptoms and a genetic mutation on *SCN1A* gene. Prolonged seizure control was obtained after using stiripentol, confirming thus the possibility of complete sustained seizure control in this epileptic syndrome.

Methods: An 8-year-10-month old boy was born to non-consanguineous healthy

parents. His parents had neither febrile nor afebrile seizures. At 5 months old, he had the first febrile seizure with generalized tonic–clonic seizures lasting a few minutes. Thereafter, he suffered various types of seizures including generalized tonic, clonic, and tonic-clonic seizures.

Results: The genetic sequence of *SCN1A* gene revealed the mutation located in exon 13 (c.2244insAACA), which caused protein translation frameshift after codon 748. On the basis of the clinical features and genetic finding, the patient was diagnosed as DS. Seizures were markedly drug-resistant and moderate mental retardation (IQ=46) was found at 7 years old. His seizures were too intractable to be controlled with a combination of valproate sodium, clonazepam and levetiracetam. At 8-year-3-month, stiripentol added led to complete control of seizures within a month, 7 years after the onset of epilepsy

Conclusion: Early recognition and diagnosis of DS and management with appropriate anticonvulsants and treatment plan may reduce the seizure burden and improve long-term developmental outcome. A prospective study is clearly needed to record the efficacy of the various anti-epileptic drugs in DS since a randomized control trial would be difficult to conduct owing to the low incidence of this epilepsy syndrome.