

Case Report:

Dravet syndrome in a 13-year-old child

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ABSTRACT

Dravet syndrome is a rare genetic epilepsy syndrome of infancy and childhood. It is characterized by occurrence of protracted febrile seizures in a normal infant followed by development of multiple seizure types and psychomotor retardation. Identifying Dravet syndrome is important, as early detection of the condition in a child presenting with febrile seizures will facilitate institution of appropriate management. We describe the rare occurrence of Dravet syndrome in a 13-year-old child who presented with mental retardation and seizures of 12 years duration.

Key words: *Dravet syndrome, India*

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INTRODUCTION

Dravet syndrome, formerly called 'severe myoclonic epilepsy of infancy, was described in 1978.^{1,2} It is classified by International League Against Epilepsy (ILAE) under Epilepsies and syndromes undetermined as generalized or focal. It accounts for 3% of epilepsy patients in first year of life.³ It begins in the first year of life, more often in first 6 months with tonic clonic seizures, almost always triggered by fever in an otherwise normal child. During the first and fourth years of life, febrile and afebrile seizures become evident. Afebrile seizures are more frequently evident as unilateral tonic-clonic or secondary generalized seizures. They evolve later to other type of seizures including myoclonic, atypical absence, atonic seizures, alternating partial seizures and convulsive status epilepticus. Early psychomotor development is normal before convulsions. Developmental stagnation is seen in the second year of life, followed by the developmental regression, accompanied by hyperactivity, language

deterioration, and mental retardation. Mutations involving sodium channel 1 alpha (SCN1A) gene are responsible for this syndrome; 80% of patients have SCN1A gene mutation.⁴

CASE REPORT

A 13-year-old female child, born to non-consanguinously married parents presented with seizures of 12 years duration. Birth history was normal. Seizures started at 9 months of age as prolonged febrile status epilepticus. Since then she had manifested multiple seizure types including febrile and afebrile generalized tonic-clonic seizures, myoclonic jerks (30 to 40 times per day) and left focal seizures with secondary generalization. She was treated with multiple drugs; partial response was observed with sodium valproate, clobazam and aggravation of seizures occurred with lamotrigene. Myoclonic seizures were severe and resistant to therapy. Her development was normal till one-and-half year of age. Subsequently her mother noticed delayed mile stones in social and language domains. She also developed

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hyperkinetic behaviour. There were no similar complaints in the family. Examination revealed severe mental retardation with a mental age of 3 years.

There were no focal motor, sensory deficits or ataxia. Laboratory investigations showed multifocal epileptiform discharges with diffuse background abnormality on electroencephalogram (EEG) (Figure 1). Magnetic resonance imaging of brain was normal.

Based on the history, examination, a clinical diagnosis of epileptic syndrome with onset during infancy with complex febrile seizures and severe myoclonic epilepsy followed by multiple seizure types during childhood was made. The differential diagnoses considered in this child were Dravet syndrome, Lennox Gestaut syndrome and Doose syndrome. On applying the screening test proposed by Hattori et al⁵, taking into account the relevant parameters when the child was under 1 year of age, a score of 7 was obtained for this child. It is recommended that if the total clinical score is 6 or more, SCN1A mutation analysis is

recommended.⁵ However, SCN1A gene mutation analysis could not be done in this patient as this investigation was not available to us and the patient's family did not have the economic resources to get it done in commercial laboratories. Based on the clinical presentation and a screening test score⁵ of greater than 6, a clinical diagnosis of Dravet syndrome was made.

DISCUSSION

Febrile seizures are the most common seizures in early childhood. The majority of febrile seizures are benign. But a small proportion of febrile seizure patients will develop epilepsy. Children with onset of seizures before one year of age, focal seizures, repeated seizures within 24 hours, prolonged seizures, and positive family history of seizures in first-degree relatives were reported to be risk factors for development of epilepsy after febrile seizures.⁶ Febrile seizure also occurs as a component in Dravet syndrome. It is an intractable epilepsy syndrome. It is difficult to differentiate the Dravet syndrome from benign febrile seizures before first year, because the clinical features

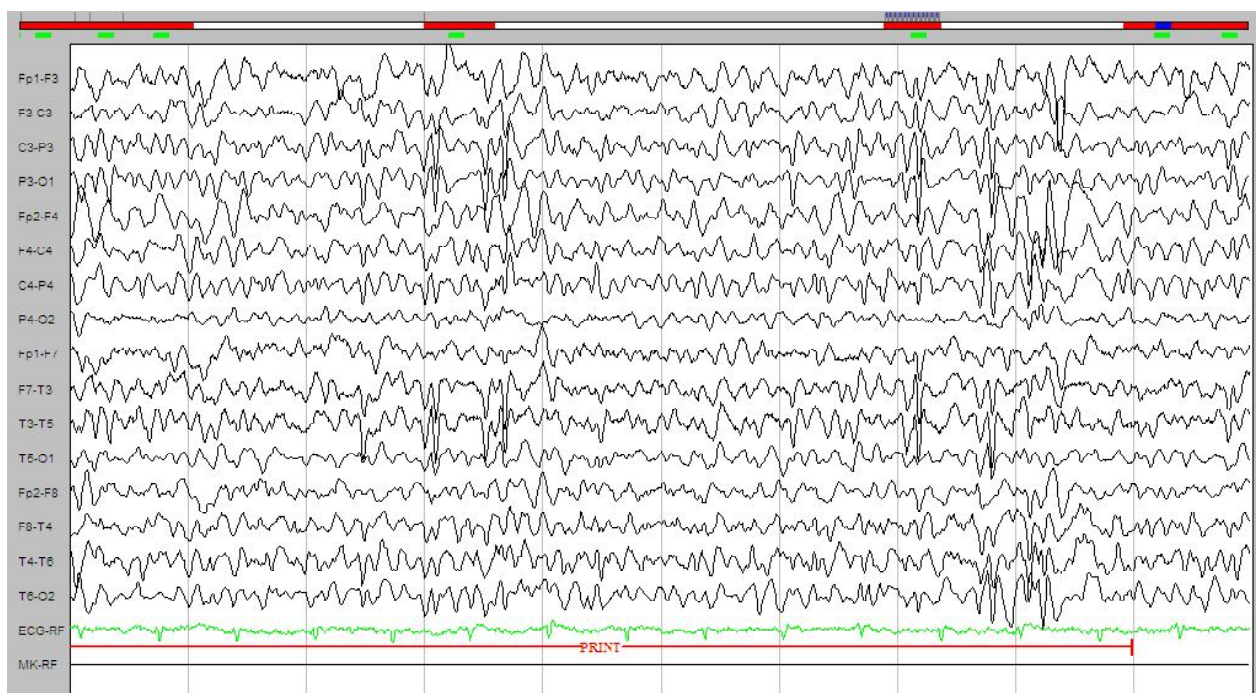


Figure 1: Electroencephalogram showing multifocal epileptiform discharges

of the syndrome are age dependent. Dravet syndrome is diagnosed after the appearance of all of its clinical manifestations at around 2 to 4 years of age.⁵ Delay in the diagnosis of the syndrome deteriorates the brain function further because of recurrent episodes of seizures which are often of prolonged duration.³

Dravet syndrome is a channelopathy resulting from a mutation of the SCN1A gene.³ This SCN1A gene mutation also results in several other epilepsies. The spectrum of diseases resulting from mutation of sodium channel gene include familial febrile seizures, generalized epilepsy with febrile seizures, intractable childhood epilepsy with generalized tonic clonic seizures, severe infantile multifocal epilepsy, severe myoclonic epilepsy (border line), and Dravet syndrome. However, the genetic test for the confirmation of diagnosis of Dravet syndrome is not available at all centres and also is expensive. Physicians should have high index of suspicion for Dravet syndrome if febrile seizures have early onset, prolonged, unilateral and concurrently have afebrile multiple types of seizures with developmental regression. Hattori et al⁵ had proposed a simple screening tool for predicting Dravet syndrome before one year of age. In

children with a score of 6 or more, SCN1A mutation analysis is recommended. A score⁵ of 7 was obtained in our patient suggesting a possible diagnosis of Dravet syndrome. There is a need for enhancing the capacity of research laboratories or evolve a network of government laboratories that can provide diagnostic facilities for rare diseases, such as SCN1A mutation analysis, free of cost to the patients.

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