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Case Report

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CASE REPORT ON DRAVET SYNDROME

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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 1 year-old child who presented with seizures which he developed at 3 months.

KEYWORDS: Dravet syndrome.

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

This 1 year old child was already diagnosed with seizure at 3 months after DPT vaccine and is on multiple anti epileptic drugs, but not adequately controlled and seizures increasing in frequency during febrile state. 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.

He has no family history of seizure. His birth history was normal. His development was normal for age. He is allergic to oxcarbazepine and now is on multiple drug therapy (C.Zonisamide 25 mg, Syp Sodium Valproate, Syp. Phenobarbitone).

Diagnosing Criteria

Dravet syndrome is a clinical diagnosis. Presentation is uniquely characteristic and, according to the 2017 consensus of North American neurologists with expertise in DS, includes:

- Typical onset between 1 and 18 months, most often <12 months, average 5.2 (Cetica 2017, Wirrell 2017).
- Recurrent generalized tonic-clonic or hemiconvulsive seizures, often prolonged but may be short
- Myoclonic seizures appearing by age 2 years, followed by obtundation status, focal seizures with impaired awareness, and atypical absence seizures.
- Hyperthermia triggers seizures in most patients (due to illness, vaccination, warm baths, exertion, etc.). Other triggers may include visual patterns or photosensitivity, eating, and bowel movements
- Normal development, neurological exam, MRI, and normal or nonspecific EEG findings at onset

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.[5]

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 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.[6] 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]

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 al[6] had proposed a simple screening tool for predicting Dravet syndrome before one year of age. Children with a score of 6 or more, SCN1A mutation analysis is recommended.

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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