Review Article

DRAVET SYNDROME/ SEVERE MYOCLONIC EPILEPSY OF INFANCY (SMEI)

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ABSTRACT

Dravet syndrome is a rare and genetic type of severe form of epilepsy characterized by episodes of prolonged, uncontrollable and progressive seizures that begin in the first year of life. It is also called myoclonic epilepsy of infancy, a rare type of epilepsy. It caused by mutations in the *SCN1A* gene and inherited from the parents in an autosomal dominant manner. Clinically features seen as frequent, prolong and refractory seizure, usually begins at first year of life and seizures that last longer than five to 30 minutes or longer. Sudden unexpected death in epilepsy (SUDEP) is more chance in this type of epilepsy. Fifteen to 20% common cause of death in sleep and also Intellectual functions(IQ) of child may be deviated. It can be diagnosed by different various investigations as EEG, MRI, history, physical examination, clinical observation, genetic testing and so on but conform diagnosis may be very late.

Key Words: *Dravet Syndrom; Myoclonic seizur; Epilepsy and Infancy*

INTRODUCTION

Dravet syndrome is a severe form of epilepsy, formerly known as severe myoclonic epilepsy of infancy (SMEI). It is rare and severe lifelong, progressive and genetic form of epilepsy. Myoclonic seizures usually begin in childhood but can occur at any age in patients with Dravet syndrome.[1] The epilepsy characterized by episodes of prolonged, uncontrolable seizures that begin in the first year of life and 80%- 85% of children survive into adulthood. It is a rare condition that affects one in 20,000 to 40,000 people worldwide. Its cause belongs to genetic condition. In around 80%–90% of patients is caused by mutations in the *SCN1A* gene (gene that encodes as a sodium channel,1.1 protein in nerve cell) and prolong repeated fever. In the remaining cases, the disease is thought to be the result of mutations in other genes, are inherited from the parents in an autosomal dominant manner, meaning that only one faulty copy of the disease-causing gene is sufficient for the condition to develop.[2,3]

Clinical features and effect on the body

Clinically it is characterized by frequent, prolong and refractory seizure, usually begins at first year of life, is characterized by seizures that last longer than 5-30 times, or longer, and are difficult to control with anti-epilepsy medications. Seizure types are usually tonic-clonic also called convulsive involving muscle jerks or other body movements. Clonic seizures consist of rhythmic jerking movements of the arms and legs, sometimes on both sides of the body that can be seen by the person observing.[1,4]

There may be whole-body convulsive seizures, such as grand mal seizures, The seizures may be triggered by fever, illness, a warm bath, or warm weather and sunlight, particularly in the early phases of the disease. More than 90% do not have control. Other problems may be associated. Sudden unexpected death in epilepsy (SUDEP) is more chance in this type of epilepsy. Fifteen to 20% cause death in sleep. Intellectual functions of child are deviated in this problem. It may range from sleeping disorders and behavioral problems to a higher risk of infections. Most children with Dravet syndrome experience developmental problems to some extent, such as cognitive impairment, and weakness in muscle coordination (or ataxia).[3]

Diagnostic procedures and possible findings

An accurate diagnosis of Dravet syndrome can be slow to arrive, because the symptoms progress with time and the initial diagnosis depends mainly on the patient's

medical history. A good description of the symptoms is the first step. Written reports of what happens from the person having the seizure and from observers can be very helpful.

History

Essential of full clinical history of the patient based on symptoms.

Information gathered by clinicians is the time the seizures started, their frequency and duration, possible triggers of seizures, and the patient's development. Genetic testing is useful to diagnose the condition and consists of a simple blood test to detect if the patient has a mutation in the *SCN1A* gene or other epilepsy-related genes. Genetic testing can be done with a simple blood test to detect mutations in the *SCN1A* gene, which are responsible for about 80 percent of all Dravet syndrome cases. These tests may also be used to investigate the presence of other epilepsy-related genes.

Electrocardiography

An EEG measures the electrical signals that the brain produces, or its activity. In Dravet patients, these signals often look normal initially, but can start showing abnormalities when the patient is between 2 and 3 years old. Abnormal signals can be single events or bursts of waves and spikes.

MRI

MRIs are usually normal at first. However, over time MRI scans may show signs of brain atrophy, abnormal growth of the cortex (the outer layer of the brain), and nerve damage in a brain region called the hippocampus (hippocampal sclerosis) in some individuals.

Other imaging methods, such as single photon emission computed tomography (SPECT) and positron emission tomography (PET) are under investigation as diagnostic tools for Dravet syndrome.

The appropriate therapeutic care for patients

Goals of managements

To terminate seizure activity

To ensure adequate systemic and cerebral oxygen delivery

To diagnosis and treat underliying disorders

To prevent complications

Previous studies have shown that treatment with sodium valproate, topiramate, or clobazam may reduce seizures in Dravet syndrome patients. These medications can be combined to reduce seizure frequency more effectively. More recently, Fintepla (fenfluramine) was approved in 2020 by U.S. Food and Drug Administration (FDA) for the treatment of seizures in patients with Dravet syndrome. Additionally, a keto-genic diet- restrictive diet low in carbohydrates, limited in protein, and high in fat may be prescribed if anti-epileptic medicines do not help the patient sufficiently. These seizures usually are often more than 5 minutes and highly resistant to many existing medications and treatments for epilepsy. There may be status epilepticus. These types of seizures are life-threatening and require urgent medical treatment, which may include oxygen, breathing support, and intravenous (into-the-vein) fluid. The sooner emergency care is given, the better the outcome.[2,4,5]

Prognosis and consequences

Dravet syndrome is a rare type of epilepsy a lifelong condition with serious implications on the quality of life of patients and their families. Long and frequent seizures may have severe consequences, including sudden unexpected death in epilepsy, status epilepticus, and a higher risk of accidents such as drowning or injuries.[5] However, with regular care and supervision, the majority of children with Dravet syndrome have a normal life expectancy. (Frequent and lasting seizures with long in duration are the most common symptoms of Dravet syndrome. These seizures usually are often more than 5 minutes - and highly resistant to many existing medications and treatments for epilepsy. While there is no cure for Dravet syndrome, early recognition can help reduce some of the problems caused by the disorder, limit the impact of other health concerns the child might have. There may decrease of cognitive function. Infections due to a weakened immune system also are possible.[6]

Nursing Management

All nursing management similar and related to epileptic seizure concerned to comfort, rest with antiepileptic medicines, prevention of injuries or accidents, control of frequent seizures and its consequences, parental counseling and all needed basic care of child suffering from this problem.[5,7]

During Seizure

First of all, stay with child, ask someone for help, protect their airway during a seizure, patients are at high risk for aspiration of their saliva, as well as position the patient. Maintain in lying position in flat surface; turn head to side during seizure activity; loosen tight clothing from neck or chest and abdominal areas;

Maintain patent airway to avoid medical emergency. Place oral airway when possible suction as needed; supervise supplemental oxygen or bag ventilation as needed. Provide supplemental oxygen. Hypoxia can not only be the cause of the seizure, but it can occur during the seizure as well.

In the event of a seizure, do not try to hold the child down or restrain them. Do not insert any objects in the child's mouth.

Maintain safety to patient, Inspect and remove potential safety hazards from the patient's surroundings. Safety is a key component of seizure nursing interventions,

Medication may be given in an attempt to stop the seizure.[4,5,6]

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