ED Management of Status Epilepticus in Pediatric Patient with Dravet Syndrome

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Dravet Syndrome is a rare, early-onset pediatric epilepsy characterized by prolonged, treatment-resistant seizures and a spectrum of neurodevelopmental delays (2). It is a sodium channelopathy, most commonly caused by a mutation in the alpha-1 subunit of voltage-gated sodium channel gene (SCN1A) (2). Dravet Syndrome is estimated to affect 1 in 15,700-40,000 live births (2), with onset of seizures typically between 1-18 months old. Diagnosis and management is challenging as there is lack of universal diagnostic guidelines and treatment protocols. Furthermore, seizures associated with Dravet Syndrome are typically refractory to medical management and routinely present in status epilepticus. The mainstay treatment algorithm for status epilepticus, using Sodium Channel blocking agents, are contraindicated in treating patients with Dravet Syndrome. It is essential for Emergency Medicine Physicians to not only recognize Dravet Syndrome, but to be familiar with appropriate treatment and management of these patients presenting in status epilepticus. This disease carries high morbidity and mortality, making quick intervention a necessity for these patients.

**Figure 1. Clinical Features of Dravet Syndrome**

- **Abnormal Seizures**: Convulsive and myoclonic seizures, primarily nocturnal.
- **Mental Retardation**: Developmental delay, intellectual disability.
- **Atypical Postures**: Seizures may begin with atypical postures.
- **Fever**: Febrile seizures can trigger status epilepticus.
- **Familial History**: Often seen in families with a history of epilepsy.

**Figure 2. OneED Management of Status Epilepticus with seizure resolution**

- IV benzodiazepine
- 0.1mg/kg (IV Valium)
- IV Levetiracetam
- 200mg/kg IV
- IV Phenobarbital
- 0.5 mg/kg
- Midazolam continuous infusion

**Discussion**

Dravet Syndrome is primarily identified by key clinical features (Figure 1) and further supported by genetic testing, although it is not necessary for diagnosis. Criteria to help guide diagnosis, treatment, and management standards was recently created by The North American Consensus Panel. Seizures associated with DS are typically resistant to pharmacologic therapy. Complete seizure control is unlikely as most patients require 2-3 anti-epileptics to achieve optimal control.

The importance of ED recognition of Dravet syndrome is that treatment regimen is unique, in that different treatment algorithms for status epileptics is contraindicated in DS (as Na channel blocking agents). For ED physicians, initial diagnosis is unlikely to take place in the Emergency Room, although you may be treating young infants on initial seizure presentation. It is important to work with local neurologist and pediatric specialists to individual seizure protocol for when these patients presents to the local ED. It is equally important for parents of affected patients to have a home regimen of rescue medications (rectal diazepam, Buccal/nasal midazolam).

**Conclusion**

- Dravet Syndrome is rare with high frequency of refractory status epileptics
- Disease carries high morbidity and mortality
- It is important for ED physicians to be familiar with disease in order to appropriately treat and avoid poor patient outcomes with use of exacerbating anti-epileptics
- Universal guidelines on in-hospital management is yet to be established. Although strong consensus that swift elimination of prolonged seizures or status epileptics is among the highest priorities in treatment.

**Tables**

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**Abstract**

Dravet Syndrome, formerly known as “severe myoclonic epilepsy of infancy” is a rare form of infantile infantile epileptic syndrome characterized by prolonged, treatment-resistant seizures and a spectrum of neurodevelopmental delays after seizure onset (2). Reports from recent studies suggest Dravet Syndrome may account for 3-5% of epilepsy cases in patients whom initial seizure presentation occurs by 3 years old (2). Pathophysiology of disease is poorly understood, but believed to be due to a mutation in alpha-1 subunit of voltage-gated sodium channel gene (SCN1A). This causes impaired action firing in GABAergic interneurons, leading to dysfunction of inhibitory interneurons (disinhibition), thus creating a hyperexcitable state.

Dravet Syndrome is typically refractory to medical management and routinely present in status epilepticus. The mainstay treatment algorithm for status epilepticus, using Sodium Channel blocking agents, are contraindicated in treating patients with Dravet Syndrome. It is essential for Emergency Medicine Physicians to not only recognize Dravet Syndrome, but to be familiar with appropriate treatment and management of these patients presenting in status epilepticus. This disease carries high morbidity and mortality, making quick intervention a necessity for these patients.

**Figure 3. Current treatment algorithm for status epilepticus in Dravet Syndrome patients**

- IV benzodiazepine
- 0.1mg/kg
- IV valproic acid
- IV levetiracetam
- IV phenobarbital
- IV midazolam

**Figure 4. Maintenance Treatment Algorithm**

- AVOID sodium channel anti-epileptics in they exacerbate seizures (phenytoin, carbamazepine, oxcarbazepine, valproate, phenobarbital, clobazam).

**References**