WHAT IS DRAVET SYNDROME?

AN INFORMATIONAL GUIDE
The goal of treatment is to reduce or eliminate seizures and to minimize their impact on development.3,4

6% of children with epilepsy under 3 years1

The acute onset of seizures in a developmentally normal infant can signify Dravet syndrome, a severe treatment-resistant epilepsy.3,5

1 in 15,700 infants2

Dravet syndrome persists throughout the patient’s life1

The prevalence of Dravet syndrome persists throughout the patient’s life1

DRAVET SYNDROME

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A rare and severe genetic epilepsy syndrome6:

• 70% to 85% of patients present with mutations in the SCN1A sodium channel gene3,6

• >100 known sodium channel mutations7

Presence of mutation without clinical signs is not sufficient for diagnosis and absence of mutation does not exclude diagnosis of Dravet.6

*May be higher with increased use of genetic testing.

AN INFORMATIONAL GUIDE
INFANCY

Seizures typically develop in the first year of life in infants who have otherwise been developing normally.\(^3,5\)

The initial seizure is often triggered by an illness and may present as a prolonged generalized or hemiclonic seizure.\(^3,5\)

Initial EEGs and MRIs are often normal.\(^3\)

DRAVET SYNDROME HAS DISTINCT CHARACTERISTICS TO CONFIRM DIAGNOSIS

**CLINICAL PRESENTATION IN INFANCY:**

**BIRTH**

The most distinctive seizure subtype is a prolonged hemiclonic seizure, although not all patients experience this seizure type.\(^3\)

Other common seizure types in the first 2 years include myoclonic and generalized tonic-clonic seizures.\(^3\)

Seizures often evolve into status epilepticus.\(^6\)

**2 YEARS**

Dravet syndrome has a broad differential diagnosis but has unique clinical characteristics to confirm a correct diagnosis\(^6,8\):

- Temperature sensitivity
- Prolonged seizures in an otherwise normally developing infant
- Developmental delays following early normal development
- Myoclonic seizures
During the second year of life\textsuperscript{3,6}: EEG abnormalities may emerge. While the child has normal cognitive development prior to seizure activity, cognitive deficits emerge between 18 months and 5 years of age.\textsuperscript{3,6} Children have moderate to severe intellectual disability and slow language progression during the second year of life.

Children frequently present to the emergency department with status epilepticus, which can lead to: \textsuperscript{5,6,11,12}

- Brain edema
- Brain herniation
- Death

**Seizures triggered by...**

- Hyperthermia
- Bathing
- Flashing lights
- Emotional stress
- Visual patterns
- Overexertion

**CLINICAL PRESENTATION AFTER AGE 2:**

- Myoclonic, focal impaired awareness, and atypical absence seizures\textsuperscript{3}
- Obtundation status\textsuperscript{3,*}

**2 YEARS**

- Impaired oculomotor coordination, dysarthria, oral motor skill deficits, impaired communication\textsuperscript{5,9}

**3 YEARS**

- Crouched gait, hypotonia, incoordination, impaired dexterity\textsuperscript{3}

**4 YEARS**

**8 YEARS**

- Lower height/weight compared with peers\textsuperscript{10}

\textsuperscript{*}Usually occurs in children older than 2 years; typically observed as nonconvulsive status epilepticus in younger children.
Seizures persist, occurring more often during sleep.\textsuperscript{5}

\textbf{Behavioral disorders are observed}\textsuperscript{5}:  
- Attention deficits/hyperactivity  
- Oppositional and personality disorders  
- Autistic traits  
- Psychotic features

\textbf{Motor signs include}\textsuperscript{3,5,8,16}:  
- Intention tremor  
- Extrapyramidal rigidity  
- Walking difficulties (including crouched gait)  
- Balance problems

\textbf{Signs and symptoms include}\textsuperscript{3,6,17}:  
- Sleep disturbance  
- Decreased response to pain  
- Dysphagia  
- Intolerance of heat and cold  
- Constipation

Sudden unexpected death in epilepsy (SUDEP) is not uncommon in children with Dravet syndrome.\textsuperscript{14}

7\% to 18\% mortality rate in patients under 18 years\textsuperscript{1,15}

SUDEP is the leading cause of death.\textsuperscript{14}

Risk factors for SUDEP include treatment-resistant generalized tonic-clonic seizures, developmental delay, and polytherapy.\textsuperscript{14}
Time is of the essence
Given the severity of seizures, including status epilepticus, and the increased risk of SUDEP in patients with Dravet syndrome, early and accurate diagnosis is urgently needed.

Increased emergency department visits to treat status epilepticus

What to do if you suspect Dravet syndrome?

Seizures persist despite standard AED treatment

Seizures may be worsened by AEDs that target sodium channels

References: