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Understanding Dravet Syndrome

A Guide for Parents



What is Dravet syndrome?

First described by French physician Dr. Charlotte Dravet in 1978, Dravet syndrome (also known as Severe Myoclonic Epilepsy of Infancy, or SMEI) is a neurodevelopmental disorder beginning in infancy and characterized by intractable seizures. Estimates of the prevalence of this rare disorder range from 1:20,000 to 1:40,000 births, but the incidence may be found to be greater as the syndrome becomes better recognized and new genetic evidence is discovered.

What is the course of Dravet syndrome?

The course of Dravet syndrome is variable from child to child. Seizures begin during the first year of life and development is normal prior to their onset. In most cases, the first seizures occur with fever and are generalized tonic-clonic (grand mal) or unilateral (one-sided) convulsions. These seizures are often prolonged and may require emergency intervention. Over the coming weeks or months, seizures increase in frequency and begin to occur without fever. Additional seizure types later appear. Most commonly, these are myoclonic, complex-partial and atypical absence seizures.

During the second to fourth year of life, varying degrees of developmental delay typically become apparent and can include regression of acquired skills. Other symptoms—such as ataxia, sleep disturbance, orthopedic issues and behavior problems—may also develop.

As children grow older, partial and myoclonic seizures may lessen, and in some cases disappear, but convulsive seizures typically persist, often occurring in sleep. Communication, motor and cognitive functions stabilize, but significant delays remain to varying degrees.

Despite being at increased risk for accident, infection and sudden unexpected death in epilepsy (SUDEP), an individual with Dravet syndrome has an 85% chance of surviving into adulthood. Because this disorder is rare and has relatively recently been identified as a distinct syndrome, little is known about long term prognosis and life expectancy.

What causes Dravet syndrome?

Identifying the causes of Dravet syndrome presents complex research problems. One known contributor is mutations of the SCN1A gene. The SCN1A gene contains instructions for the creation of proteins that regulate the function of sodium ion channels within the brain. A mutation of this gene may lead to improper functioning of the sodium ion channels, incurring seizures. Roughly 70% to 90% of patients with a clinical picture of Dravet syndrome will test positive for an SCN1A mutation.



Researchers have documented many different mutations of the SCN1A gene, however most of them do not result in Dravet syndrome. SCN1A mutations more often lead to milder forms of epilepsy, including Generalized Epilepsy with Febrile Seizures (GEFS), Generalized Epilepsy with Febrile Seizures Plus (GEFS+), Intractable Childhood Epilepsy with Generalized Tonic-clonic Seizures (ICEGTC) and SME Borderline (SMEB). Dravet syndrome is generally considered to be the severe end of a broad spectrum of SCN1A-related epilepsies.

Mutations of the SCN1A gene can be passed from parent to child. However, in Dravet syndrome, while many of the affected individuals have some history of febrile seizures or epilepsy in their extended family, the gene mutation nearly always arises 'de novo', or new to the individual. Much remains to be understood about the causes of Dravet syndrome and research is ongoing.

Currently, treatment for Dravet syndrome consists mainly of anticonvulsant medications to help control seizures. Response to these medicines is variable, but often the seizures persist despite treatment. While certain medications have been found to be generally useful for individuals with Dravet syndrome, others have been quite consistently found to have an aggravating effect. The helpfulness of other types of anticonvulsant therapies, such as the ketogenic diet, vagus nerve stimulation (VNS), and IVIg therapy are in ongoing evaluation.

What is the IDEA League?

The International Dravet syndrome Epilepsy Action League is an international partnership of parents and professionals united in the purpose of creating greater awareness and understanding of Dravet syndrome and related conditions. The IDEA League was founded by parents in October of 2005.

Our **mission** is to promote awareness of and research for Dravet syndrome and related forms of epilepsy and provide resources and support to improve the quality of life for affected individuals and families. We strive to be the most up-to-date and accurate information source for Dravet syndrome and related conditions for both families and professionals.

You can help! Because the IDEA League is a non-profit, charitable organization, support from the community and those with an interest in Dravet syndrome is crucial for us to achieve our goals. Please consider joining the League, volunteering time or resources, or providing financial support. Donations are tax-deductible within the USA.

While based in the United States, the IDEA League is an international organization comprised of hundreds of families, as well as leading physicians and scientists, from around the world. We provide services to members on every inhabited continent, and are working to meet the needs of all our membership with affiliate organizations in the United Kingdom and France, with others in development. Joining the IDEA League allows you to be part of a global team working to improve the outcomes of children with Dravet syndrome and related epilepsies.

Our IDEAL is a Cure. Together, we will find it!

To join the IDEA League or to make a donation, complete and mail the form on the reverse of this pamphlet or visit us online at www.IDEA-League.org. Thank you.