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Please use my donation for ☐ research or ☐ family support-awareness (optional)

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DRAVET Syndrome UK

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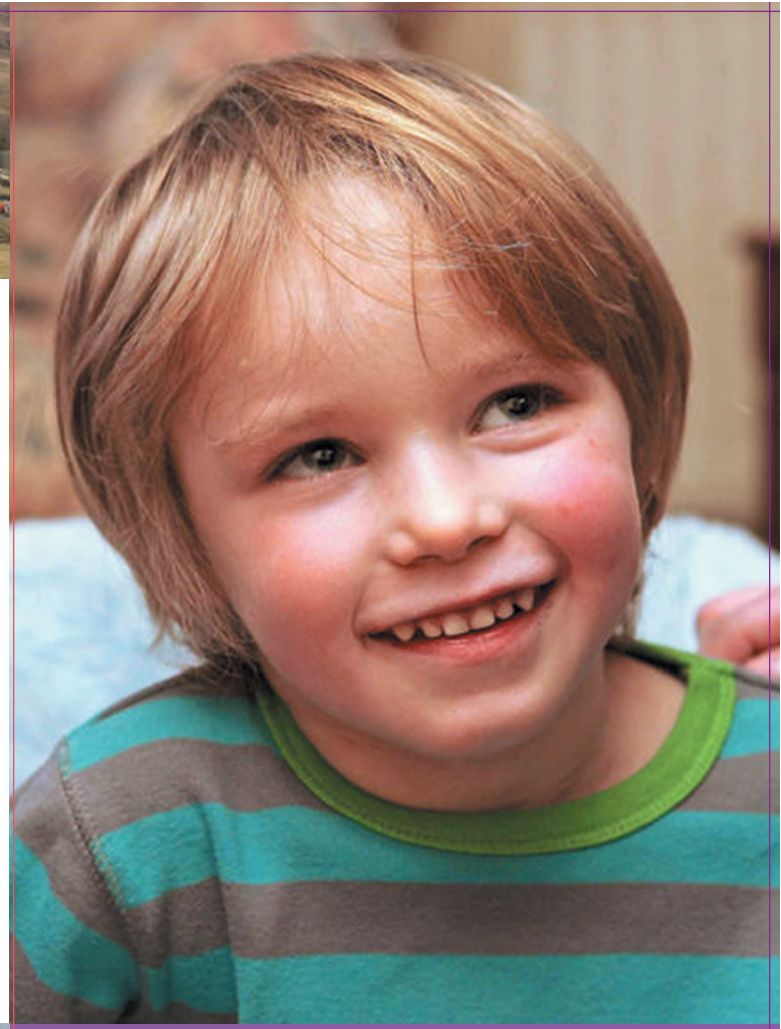
DRAVET Syndrome UK

Promoting awareness of Sodium Channel Epilepsies



Understanding Dravet Syndrome

DRAVET Syndrome UK

What is Dravet syndrome?

First described by French physician Dr. Charlotte Dravet in 1978, Dravet syndrome (previously known as Severe Myoclonic Epilepsy of Infancy, or SMEI) is a neurodevelopmental disorder beginning in infancy, characterised 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 recognised and new genetic evidence is discovered.

What is the course of Dravet syndrome?

The course of Dravet syndrome is variable from one child to another. Seizures typically begin during the first year of life and neuro-development is normal prior to their onset. In most cases, the first seizures occur with fever and are generalized tonic-clonic (involving the whole body) or one-sided convulsions. These seizures are often prolonged (lasting longer than 5 minutes) and may require emergency intervention. Over the next weeks or months, seizures increase in frequency and begin to occur without fever. Additional seizure types later appear. Most commonly, these are myoclonic (single jerks), focal or atypical absence seizures (usually involving staring and unresponsiveness). During the second to fourth year of life, varying degrees of developmental delay typically become apparent. Other symptoms – such as ataxia (unsteadiness), sleep disturbance, and behaviour problems - may also develop. As children grow older, focal and myoclonic seizures may lessen, and in some cases disappear, but convulsive seizures typically persist, often occurring from sleep. Prolonged seizures continue to be a risk and may still be more likely to occur with fever or illness. Communication, motor and cognitive functions stabilise, but significant delays persist.

Previously children with an apparently milder clinical course, usually without myoclonic seizures, have been described as having severe myoclonic epilepsy borderline (SMEB). It is now accepted to classify these children as also having Dravet syndrome.

Children with Dravet syndrome are at a higher risk of sudden unexplained death in epilepsy (SUDEP) than children with other types of epilepsy. Despite this they have an 85% chance of surviving into adulthood. Adults are increasingly being recognised with this type of epilepsy as adult specialists become more aware of the condition.

What causes Dravet syndrome?

A major contributor to the cause of Dravet syndrome has been found to be mutations of the SCN1A gene, found in around 80% of patients. This gene contains instructions for the creation of proteins that regulate the function of sodium ion channels. A change (mutation) in this gene may lead to abnormal functioning of the sodium ion channels in the brain, which are presumed to cause seizures.

Researchers have documented many different mutations of the SCN1A gene. However, not all of these result in Dravet syndrome. SCN1A mutations are also associated with other forms of epilepsy, including Intractable Childhood Epilepsy with Generalized Tonic-clonic Seizures (ICEGTC) and Severe Multifocal Epilepsy of Infancy.

In Dravet syndrome, the gene mutation nearly always arises 'de novo', or new to the individual, even though many of the affected individuals have some history of febrile seizures or epilepsy in their extended family, Rarely, mutations of the SCN1A gene, can be passed from parent to child. Much remains to be understood about the causes of Dravet syndrome and research is ongoing. In those children negative for SCN1A mutations, other genetic mutations are being explored.

Treatment of Dravet Syndrome

Currently, treatment for Dravet syndrome consists mainly of anti-epileptic medications to help control seizures. Response is variable, but often the seizures persist despite treatment. Certain medications – currently including sodium valproate, clobazam, stiripentol, topiramate, levetiracetam - have been found to be generally useful. Others have been found to have an aggravating effect (eg lamotrigine, carbamazepine). The ketogenic diet has also been shown to be particularly useful in some individuals. The role of vagal nerve stimulation (VNS) still requires evaluation.

Medical information written by Professor Helen Cross,
UCL - Institute of Child Health,
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DRAVET Syndrome UK

DRAVET Syndrome UK is a registered charity dedicated to improving the lives of those affected by Dravet Syndrome in the United Kingdom. DRAVET Syndrome UK was established in October 2008 and registered as a UK charity in February 2009.

DRAVET Syndrome UK has 3 main aims:

1. To fund medical research into Dravet Syndrome and other related genetic sodium channel epilepsies.
2. To advance the education of the medical community in the UK with regards to Dravet Syndrome, therefore prompting accurate and early diagnosis.
3. To promote the physical and mental health of sufferers of Dravet Syndrome and their families within the UK through the provision of financial assistance, support, education and practical advice.

How you can help:

Because DRAVET Syndrome UK is a non-profit, charitable organisation, support from the community and those with an interest in Dravet Syndrome is crucial in helping us achieve our aims.

Donations from UK taxpayers are eligible for Gift-Aid. You can also donate securely at www.justgiving.com/dravetsyndromeuk

Please help us work towards a cure for Dravet Syndrome.

To join DRAVET Syndrome UK or to make a donation please fill in the form on the reverse of this leaflet and post it to the address given. Alternatively you can visit us online at: www.dravet.org.uk

Many thanks.