



## What is a syndrome?

A syndrome is a group of signs and symptoms which, if they occur together, can suggest a particular condition.

Many children and young people will have a set of related symptoms which fit a particular pattern and this will determine which syndrome they have.

Knowing which syndrome will help the doctors to choose the appropriate antiepileptic drug (if needed) and also give a more accurate prognosis for the child/young person. It can also help to point parents in the right direction to get help and support.

Knowing the syndrome will be useful for understanding:

- whether the child/young person's seizures are likely to be controlled
- which medication is likely to work best
- whether there are likely to be any other problems, for example with behaviour, learning or social functioning
- long term prognosis

## Dravet Syndrome

Dravet syndrome is an extremely rare epilepsy syndrome.

### Key features include:

- onset of frequent and prolonged febrile convulsions in response to a high temperature in the first year of life;
- development of other seizure types in the second year of life
- Episodes of status epilepticus (prolonged seizures)
- Seizures do not respond well to antiepileptic drugs
- severe learning difficulties
- developmental decline and loss of skills

## Seizures

In the first year of life, infants frequently experience prolonged febrile convulsions (usually lasting 15-30 minutes or longer) in response to a high temperature. These febrile convulsions are either tonic-clonic or clonic in nature and may affect typically one or at times both sides of the body.

- **Tonic clonic seizures** – when these seizures occur there is a loss of consciousness resulting in the child going stiff and falling. This is followed by repetitive jerking of the limbs
- **Clonic seizures** – when these seizures occur there is a loss of consciousness resulting in the child

falling. This is followed by repetitive jerking of the limbs.

In the second year of life, seizures become more frequent. In addition to tonic-clonic and/or clonic seizures, other seizure types may develop including focal and myoclonic seizures and absences. Although these seizures can occur at any time, high temperatures and flashing lights are identified seizure triggers for many children with Dravet Syndrome

- **Focal seizures** – these seizures come from one area of the brain; what the seizure looks like depends on the function of that area.
- **Myoclonic seizures** – the child has sudden muscle jerks. Seizures usually become less frequent once the child enters their early teens.

## Causes

80% of children with Dravet Syndrome have a genetic mutation (damage to their genetic material) in a specific section called SCN1A. In the majority of cases, this mutation arises spontaneously in the child when they are an embryo, rather than being passed down through the family.

For the other 20% whom do not have the SCN1A mutation, the cause of their epilepsy is largely unknown, although many other possible genetic mutations are being explored.

## Diagnosis

Diagnosis of Dravet syndrome is based on several factors, including:

- **Clinical history:** A full history of the child's seizures, age of onset and development is vital.
- **EEG findings** (a non-invasive painless test that records brain activity by picking up electrical signals given off by nerve cells): Whilst EEG results are often normal within the first year of life, abnormalities may become evident from approximately 18 months.
- **A genetic test:** 80% of children with Dravet Syndrome have a genetic mutation (damage to their genetic material) in a specific section called SCN1A. A blood test can be used to confirm whether or not the child suspected of having Dravet Syndrome has the SCN1A mutation or not. However, 20% of children do not have this mutation and therefore its absence does not rule out the diagnosis.

## Treatments

In many cases Dravet Syndrome proves resistant to antiepileptic medication, even when additional drugs in high doses are added to the treatment regime (polytherapy). The antiepileptic drugs most frequently used are sodium valproate, clobazam, topiramate, levetiracetam and stiripentol.

In cases where antiepileptic medication is having little or no effect, the Ketogenic diet may be considered. This is a high fat, low carbohydrate and controlled protein diet that requires strict supervision and even the slightest deviation from the prescribed diet may cause the diet to lose its effect.

## Outcome

Although seizure control remains poor throughout childhood, seizure frequency often improves in the early teens. However, children and young people with Dravet Syndrome frequently have severe learning difficulties and developmental delay – particularly in the areas of speech and language – that persist despite less seizures. A typical gait may be seen later. As a result, those with Dravet Syndrome will require care and support from others throughout their lives.

## For information and support:

Contact a Family  
[www.cafamily.org.uk](http://www.cafamily.org.uk) or ring 0808 808 3555.

## Young Epilepsy Helpline

If you would like to know more about epilepsy, treatments, causes or for general information about medication – we are here to answer your questions. Talk privately with our experienced team in complete confidence, we can also provide information and support.

Simply contact us on:

**Phone:** 01342 831842, from 9am – 1pm, Monday to Friday.

**Email:** [helpline@youngepilepsy.org.uk](mailto:helpline@youngepilepsy.org.uk)

**Text:** 07860 023 789, texts are charged at your standard rate.