

## 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

## Aicardi Syndrome

Aicardi syndrome is an extremely rare neurological disorder that almost exclusively affects girls.

Many girls affected by this syndrome will have:

- an absence or partial absence of a structure within the brain called the corpus callosum that connects the two halves of the brain;
- infantile spasms in early infancy;
- and defects in the light-sensitive tissue of the eye (the retina).

Other features of the condition may include:

- additional eye defects;
- a small head;
- a cleft lip;
- curvature of the back;
- and/or gastrointestinal difficulties.

## Seizures

Infantile spasms often occur within the first three months of life. Over time, seizures become more frequent and difficult to control. Some children may go on to also experience focal seizures.

## The causes

Aicardi syndrome is a genetically inherited condition. However, it is not passed down through families but is instead caused by a random change (mutation) in the child's genetic makeup at the point of conception.

## Diagnosing Aicardi Syndrome

Diagnosis of Aicardi syndrome is based on several factors including:

- the nature of the child's seizures and their history;
- the results of investigations, such as MRI brain scans and an EEG (a non-invasive painless test that record brain activity by picking up electrical signals given off by nerve cells);
- and the outcome of an eye examination.

## Treatment

Antiepileptic drugs that may be useful include vigabatrin and sodium valproate and in some cases a course of corticosteroids may be beneficial. However, seizures are often resistant to medication and therefore alternative treatments such as the Ketogenic Diet may be considered.

## Prognosis

The majority of girls with Aicardi syndrome will have moderate to severe learning difficulties and will remain dependent on others for the duration of their life. Some

girls may have normal vision, whereas others may be blind or partially blind. A minority of girls may develop some language skills and aided mobility.

## For information and support:

Contact a Family  
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.