

## 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 that fit a particular pattern and this will determine which syndrome they have.

Identifying the 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

## Sturge Weber syndrome

Sturge-Weber (SWS) is a rare condition that occurs early in pregnancy at around 6-9 weeks gestation. It is not a genetic condition that is inherited and it is unknown as to why it occurs.

Children with SWS may have associated conditions such as epilepsy, hemiplegia, visual field defects, headaches, glaucoma and developmental delay and learning difficulties.

### Port wine birth mark

It is thought that a network of blood vessels continues to develop in utero causing an extra layer of blood vessels over the surface of the brain called an angioma.

The abnormality in the network of blood vessels results in a port wine coloured birth mark on the face around the forehead and/or scalp.

### Epilepsy

In over two thirds of cases children with SWS that develop epilepsy. The seizures may start at around birth and usually before the age of 2 years. The onset of epilepsy is often associated with the child having a high temperature.

## Better futures for young lives with epilepsy

The seizure types can vary and often occur as jerks on one side of the body (focal seizures). Some may then generalise and include other seizure types. The seizures may occur frequently and, in some cases, may be prolonged.

### Hemiplegia

As a result of the angioma, some children with SWS will have weakness in the opposite side of the body from the side with the port wine stain. The degree and onset of the weakness is variable from child to child and may develop in infancy, along with seizures, or occur in association with the headaches and in relation to the frequency and severity of the seizures.

### Visual Field Defects

Some children with SWS have a visual field deficit. This means that they will have difficulty seeing objects out of the corner of their eye.

### Headaches

Around a third of children with SWS may suffer from migraine or severe headaches which are often associated with seizures.

### Glaucoma

The raised pressure in the abnormal blood vessels around the eye can result in a child with SWS suffering from increased pressure within the eye. This is known as glaucoma. This may be present at birth, but the majority of children develop it in infancy, although for others it may be later.

For this reason all children with SWS

should have the pressure within the eye checked regularly.

### Developmental delay

About 60% of children with SWS will have developmental delay and some level of learning difficulties. Children with abnormal blood vessels on both sides of the brain are more likely to have more severe learning disabilities. Those children who develop epilepsy are also more likely to have developmental delay.

### Diagnosis

The presence of a deep red birth mark on the forehead or scalp alerts doctors to the possible diagnosis of SWS. An MRI scan after the child has been given an injection of a dye called gadolinium to make the blood vessels show up better will confirm the diagnosis. A mutation in the GNAQ gene has been identified in many patients.

### Treatment

Since SWS has a number of symptoms; a variety of professions will be involved in the child's treatment. Specialist involvement will include neurologists, dermatologists, neurologists, ophthalmologists and child development team.

### Port wine birth mark

Because port wine stains on the face may have an emotional or social impact on a child, input from a psychologist might be required. More and more children have had their birth marks successfully treated with lasers.

## Epilepsy

Antiepileptic drugs will be the first line of treatment for a child's epilepsy. Very often different treatment regimes will need to be tried to find the optimum treatment for the seizures. Some children will struggle to get seizure control and if this is the case, epilepsy surgery or vagus nerve stimulation may be an option.

## Hemiplegia

Physiotherapy input and an appropriate exercise regime will be of great benefit to a child who experiences episodes of weakness or hemiplegia.

An occupational therapist will be able to offer advice on equipment and ways to help with tasks that are problematical for the child.

Some doctors prescribe a daily dose of aspirin which is thought to reduce the 'stickiness' of the blood and thereby is useful in reducing the episodes of severe weakness.

## Visual Field Defects

Depending on the extent of the visual field defect an ophthalmologist will be able to give advice on ways of lessening the impact on a child, for example putting objects such as toys and games clearly within the child's field of vision will help overcome these difficulties.

If the defect is very severe, specialist teachers for the visually impaired may need to be consulted.

## Glaucoma

Regular checks by an ophthalmologist will diagnose glaucoma. If this condition is present it can usually be effectively treated with regular use of eye drops. Very occasionally an operation may be required.

## Developmental delay

For those children with learning difficulties a statement of special educational needs and an individual educational plan (IEP) will be required which will detail any extra support that will be required.

## Prognosis

The prognosis for a child with SWS is very variable and is dependent on to what extent the brain is affected and on the frequency and severity of the seizures. Those children who gain good seizure control are likely to have better outcomes than those who have frequent and prolonged seizures.

## For more information and support

### **Sturge-Weber Foundation (UK)**

Burleigh, 348 Pinhoe Road Exeter EX4 8AF

Tel: 01392 464675

Website: [www.sturgeweber.org.uk](http://www.sturgeweber.org.uk)

### **Contact a Family**

Tel: 0808 808 3555

Website: [www.cafamily.org.uk](http://www.cafamily.org.uk)