Sturge-Weber syndrome

Information for families

Great Ormond Street Hospital for Children
NHS Foundation Trust

This leaflet explains about Sturge-Weber syndrome (SWS), how it can be treated and what to expect when you come to Great Ormond Street Hospital (GOSH) for diagnosis and treatment.

What is Sturge-Weber syndrome?

A syndrome is a collection of signs that are often seen together. Sturge-Weber syndrome (SWS) is a condition affecting the skin, brain and eyes. It is named after the doctors who described it in the late 19th century and early 20th century.

What are the signs and symptoms of SWS?

They affect the skin and brain as follows:

- A port wine stain is a vascular birthmark caused by abnormal development of blood vessels in the skin. It can occur anywhere on the body, but in SWS it affects the skin around the forehead and/or scalp.
- As well as the port wine stain affecting the skin, it will also involve an extra layer of blood vessels over the surface of the brain (angioma).
- The angioma may be associated with seizures (fits or convulsions) and developmental problems.

How is it diagnosed?

If your child's port wine stain is on the forehead or scalp, he or she will need to be checked by a paediatrician (specialist children's doctor). Your child will have an MRI scan with gadolinium to confirm the diagnosis. This is basically the same as a regular MRI scan, but your child will have an injection of dye (gadolinium) beforehand. This makes your child's blood vessels show up better on the scan. Once the diagnosis has been confirmed your child's care will be shared between the paediatrician and a paediatric neurologist (brain and nervous system doctor).

What causes SWS?

SWS occurs sporadically. It is not a genetic condition and so is not passed from parent to child. The condition starts very early in pregnancy when the baby is developing in the womb. At six to nine weeks of pregnancy, the tissues that will eventually form the skin and brain are closely related. We think that a network of blood vessels continues to develop instead of separating, causing an extra layer of blood vessels over the surface of the brain. We do not know exactly why this happens, but it is very unlikely that it is due to anything you did or did not do during pregnancy.

How common is it?

We do not know how many people are diagnosed with SWS but we know it is a rare disorder. It can affect all ethnic groups and is equally common in boys and girls. Children with a port wine stain on both sides of their scalp or forehead are more likely to have SWS.

What problems can the symptoms cause?

Port wine stain

Port wine stains, particularly if they are on the face, can affect children psychologically, emotionally and socially. Our leaflet *Bringing up a child whose face looks different* contains lots of ideas and suggestions from the Birthmark team and parents of children with birthmarks.

Developmental problems and learning disability

About 60 per cent of children with SWS have delayed developmental milestones and learning problems. That is, they do not develop and acquire key skills at the same age as children without SWS. This is more likely to happen in children who have SWS with epilepsy. However, children vary enormously and so children with SWS can have abilities within the normal range or have more delayed development. It seems that children with abnormal blood vessels on both sides of the brain have more severe learning disabilities than others.

Visual field deficit

Some children with SWS have a visual field deficit. This means that they have trouble seeing objects out of the corner of their eye. In infants, this can be diagnosed by clinical examination, but proper diagnostic tests are more difficult until children are of primary school age and are more cooperative. Once you know which side is affected, you can make sure that toys, games or work are put within your child's field of vision. Children with a visual field deficit may seem to use the affected side less than the other, so ways of improving this might be suggested.

Hemiplegia

Children with SWS might have a weakness in the opposite side of the body to the port wine stain due to abnormal blood vessels in the brain. They may also have difficulty using the affected side because they are less aware of it. The degree of weakness varies from child to child and may be barely noticeable or more obvious. There seem to be

three ways weakness can affect children with SWS: in one group, the weakness is noticeable during infancy, in the second the weakness develops with the seizures and in the final group the weakness may develop in association with headaches, and may improve.

Epilepsy

Around 80 per cent of children with SWS have epilepsy (fits or convulsions) if the SWS is on both sides of the face. In most of these children it starts before the age of two, and by the time a child is five years old the majority of children who are going to develop epilepsy have done so. The onset of these seizures is often triggered by an illness causing a high temperature. Various types of seizures might occur, most commonly jerky movements affecting one side of the body, although other types have been reported. If the seizures last for more than 30 minutes or happen very frequently, they may affect a child's development. Treatment is usually adjusted to reduce the frequency or severity of the seizures.

Headaches

About one third of children with SWS have headaches and migraine-like episodes. It is likely that these are caused by the blood flowing through the extra layer of blood vessels. Headaches seem to be linked to period of seizures and weakness in many children.

Glaucoma

Another feature of SWS is glaucoma. Glaucoma is raised pressure within the eye, which can lead to blindness if it is not treated. Glaucoma is diagnosed using a test that measures the

pressure in the eye by blowing a puff of air. The majority of children develop glaucoma in infancy, but some do not develop it until later childhood. The specialist eye doctor (ophthalmologist) should examine your child's eyes to check for glaucoma regularly: every year for the first five years, and then every two years. If your child develops glaucoma, he or she will need more frequent appointments.

The condition itself cannot be treated, but there are various options for treating the symptoms. As SWS involves different parts of the body, a multidisciplinary team often provides the best care. Regular reviews with your local paediatrician and child development centre alongside doctors from a specialist hospital will be needed. The specialists will usually involve dermatologists (skin doctors), neurologists (brain and nervous system doctors) and ophthalmologists (eye doctors).

SWS is a lifelong condition, needing ongoing care and treatment, so a child will need to transfer to adult services when he or she reaches 16 years old or so. We will work with you and your child to make sure that this transfer happens as smoothly as possible.

The symptoms of SWS can be treated as follows:

Port wine stain

Laser treatment, with a pulsed dye laser, is currently the treatment of choice for fading a port wine stain. When laser treatment is carried out at a specialist centre experienced in dealing with vascular birthmarks in children, the results can be excellent and the side effects minimal. With newer, up-to-date lasers these children (about 10%) who have not responded to the previous lasers have shown very promising results. For more information, please see our *Port wine stains* leaflet.

Developmental problems and learning disability

Support from therapists based at a child development centre or special needs school can be invaluable. Children with developmental problems and learning disability may also need support in school. An individual education plan (IEP) and statement of special educational need (Statementing) may be needed, as they set out the support your child needs and guarantees the extra resources for it. Some children develop thyroid problems after the age of seven or eight years, so they may require a blood test and possibly life long thryoid medicines.

Visual field deficit

The specialist eye doctor in your multidisciplinary team may be able to suggest ways of dealing with any visual field deficit. As your child grows older, certain games and sports might prove problematic, but there are plenty of alternatives or ways to get around the visual field deficit. Your child may benefit from support from a specialist teacher of children with visual impairment.

Hemiplegia

Input from a physiotherapist and occupational therapist can improve the way your child copes with the weakness. They may suggest exercise programmes to strengthen the weaker limbs or aids to make everyday tasks easier. There is some evidence that taking a low dose of aspirin every day can reduce how 'sticky' the blood is, and therefore reduce the number of episodes of severe weakness. Aspirin has been linked with a severe liver illness (Reye's syndrome) in the past, although this is very rare. You should discuss whether or not to use aspirin with your doctor.

Epilepsy

There are many anti-epilepsy medicines suitable for infants and children. It can take time to find the best combination and the ideal dosage for your child, so it is possible that your child's epilepsy treatment plan could change quite frequently.

If the seizures are still severe after trying various combinations and dosages of medicines, epilepsy

surgery might be considered. You will need an appointment at a specialist hospital carrying out children's epilepsy surgery. The assessment process for epilepsy surgery is complex, and is explained in more detail in our *Surgery for epilepsy* leaflet.

Glaucoma

Your child will have his or her eyes checked regularly so that any glaucoma can be diagnosed and treated quickly. Treatment is usually with eye drops and occasionally an operation.

What is the outlook for children with SWS?

Children with SWS have a range of disability, from mildly affected to severe. The outlook varies greatly from child to child, depending on the severity of the symptoms and how well they are controlled.

Is there a support group?

Yes, the following organisation can offer you help, support and advice.

Sturge-Weber Foundation (UK)
Burleigh, 348 Pinhoe Road
Exeter EX4 8AF

Tel: 01392 464675
Website: www.sturgeweber.org.uk

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Compiled by the Birthmark Unit and Neurology department with assistance from parents of children with SWS in collaboration with the Child and Family Information Group

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