



# PHACES association

This information sheet from Great Ormond Street Hospital (GOSH) provides information about the medical condition PHACES association (previously referred to as PHACES syndrome) and what to expect when your child comes to GOSH for assessment and treatment.

## What is PHACES association?

PHACES association is the name given to a collection of features that are often seen together. PHACES stands for:

- Posterior fossa (area of brain) abnormalities
- Haemangioma
- Arterial lesions
- Cardiac abnormalities
- Eye problems
- Sternal notch or dimple

PHACES association is a spectrum disorder, that is, the effects can range from mild to severe. Most children only have one additional problem other than the haemangioma. PHACES association appears to be more common in girls than boys, although we do not yet know why this should be the case.

## What causes PHACES association?

The term PHACES syndrome was first described in 1996. Much more research is needed to understand what causes the condition and why it is more common in girls. It may have a genetic basis, but no inherited cases passed on from parent to child have been reported to date.

## What are the signs and symptoms of PHACES association?

Usually the first visible sign is a large relatively flat (plaque) haemangioma that appears on the face within a couple of weeks of birth. Some children may have additional haemangiomas elsewhere on the body. If the haemangioma affects midline structures, such as the front of the neck, jaw or lip, there is a possibility of a haemangioma affecting the windpipe.

### Posterior fossa abnormalities

Some children with PHACES syndrome have an abnormality of the part of the brain called the cerebellum. These abnormalities may have no adverse effect on the child, but may rarely lead to disrupted cerebrospinal fluid (CSF) flow around the brain and abnormalities in the area between the two hemispheres (halves) of the cerebellum (vermis). These problems may cause developmental delay, raised pressure within the brain and increased skull size, sometimes leading to problems with balance and coordination (ataxia).

### Arterial lesions

Blood flow to the brain through the internal carotid and cerebral arteries may also be abnormal in children with PHACES association. Sections of the arteries may be wider than normal, or the vessel may have additional branches or missing connections, or follow an abnormal route. In some cases, arteries may have sections that are narrower

than usual, occasionally leading to reduced blood flow to the brain. The carotid arteries may very rarely be absent on the side of the haemangioma.

### **Cardiac abnormalities**

A common cardiac abnormality seen in children with PHACES association is coarctation of the aorta. The aorta is the main blood vessel that carries blood away from the heart. The word coarctation means that a section is narrowed, leading to reduced blood flow to the lower part of the body.

### **Eye problems**

Eye problems in PHACES syndrome tend to arise on the same side as the haemangioma. If the haemangioma obscures the eye, this can lead to vision problems, such as lazy eye. Very occasionally the eye may be small (microphthalmia) or the optic nerve (which transfers images from the eye to the brain) may be underdeveloped.

### **Sternal abnormalities and abdominal raphe**

The sternum (breastbone) can be affected in children with PHACES association, having a notch or cleft or being partially or entirely absent. Even less frequent is a scar-like line extending down the front of the abdomen ('abdominal raphe').

## **How is PHACES association diagnosed?**

Usually the first sign is the haemangioma, which begins to appear shortly after birth. Children with segmental facial haemangiomas should be reviewed by a specialist paediatric dermatology (skin) doctor who will arrange for review by other specialists as required. If PHACES association is suspected, they will carry out a full physical examination, and investigation which may include magnetic resonance imaging (MRI) brain scan and cardiac ECHO to identify any brain or heart abnormalities. An injection of contrast (a liquid that shows up well on scans) will be used to map blood flow to the brain. An ECHO (echocardiogram) or ultrasound of the heart shows abnormalities of structure or blood flow. Specialist eye examination is used to diagnose eye problems or visual impairment.

## **How is PHACES association treated?**

As PHACES association affects a variety of body systems, a multidisciplinary approach involving different specialists is required. The team may include dermatologists (skin specialists), neurologists (brain specialists), cardiologists (heart specialists), ear nose and throat surgeons and ophthalmologists (eye specialists).

### **Posterior fossa abnormalities**

Not all brain abnormalities can be treated, and many do not require treatment, but any developmental delay resulting from them can be helped with the right support. If a child has abnormal CSF flow, a shunt to drain away excess fluid may be suggested.

### **Haemangioma**

Most haemangiomas do not require any treatment, but there are circumstances when treatment might be needed. Haemangiomas near the eye can have long-term effects on a child's vision, so need to be checked by a specialist eye doctor (ophthalmologist). Haemangiomas that are blocking vision may need treatment with beta blocker given as a liquid by mouth.

Occasionally, haemangiomas can form an open sore or ulcer, which is painful. If your child's haemangioma develops an ulcer, it will need special attention until it heals. Haemangiomas on the lips often become ulcerated and because ulcers are so painful, the child may not want to feed. Giving your child pain relief before feeding can help. It can also help to put some petroleum jelly on the teat of the bottle (or around your nipple if you are breast feeding) to reduce friction and make feeding less painful.

Haemangiomas on the jaw, chin or front of the neck can sometimes be associated with haemangioma in the airway, leading to breathing difficulties. The first sign of this is stridor, a rasping sound with each inward breath. This may need treatment with beta blockers (usually propranolol) or laser treatment of the airway haemangioma during a microlaryngobronchoscopy (MLB). Very occasionally a child may need to have a tracheostomy (artificial opening into the windpipe) to improve their breathing.

### **Arterial lesions**

Very rarely surgery may be suggested to widen a narrowed section of artery.

## Cardiac abnormalities

Cardiac problems such as coarctation of the aorta will need to be corrected surgically soon after diagnosis.

## Eye problems

The treatment required will depend on the nature of the eye problem. In the rare cases leading to significant reduction in vision, support from experts in visual impairment can help children to make the most of what vision they have so that they can attend school and take part in social activities.

## Sternum problems

If the sternum is partially or completely absent, surgery to repair or construct the sternum will be required early in infancy, to improve breathing problems and to ensure that the sternum and ribs form a protective 'cage' around the heart and lungs.

## What happens next?

Most haemangiomas will have disappeared completely by the age of five to seven years. Large haemangiomas may continue to get smaller until your child is about eight to ten years old. Depending on the size and location of the haemangioma, there may be little sign it ever existed. Occasionally the affected area of skin might be a bit lighter in colour than the rest of your child's skin. Remaining tiny red veins (thread veins) on the surface of the skin can be treated with a laser later on in childhood. It is possible that large haemangiomas may distort the surrounding skin and even when they disappear, leave behind an area of stretched skin that looks puckered or wrinkled. This can often be improved with plastic surgery. Some children may have an uneven skin texture once the haemangioma has resolved. Some children may have some leftover fatty tissue. This can be removed by a plastic surgeon. Haemangiomas affecting the ear or nose may leave some distortion, which can usually be improved through plastic surgery.

Although the haemangioma disappears in childhood, some children with PHACES association may continue to need regular check-up appointments to monitor any other problems throughout childhood and into adulthood.

## Further information and support

The Birthmark Support Group is the main organisation in the UK offering support and advice. Email them at [info@birthmarksupportgroup.org.uk](mailto:info@birthmarksupportgroup.org.uk) or visit their website at [www.birthmarksupportgroup.org.uk](http://www.birthmarksupportgroup.org.uk)

Changing Faces supports anyone affected by a visible difference. Call them on 0845 4500 275 or visit their website at [www.changingfaces.org.uk](http://www.changingfaces.org.uk).