PATIENT INFORMATION LEAFLET

BLUE RUBBER BLEB NAEVUS SYNDROME

WHAT ARE THE AIMS OF THIS LEAFLET?

This leaflet has been written to help you understand more about blue rubber bleb naevus syndrome. It explains what this condition is, what causes it, what you can do about it, and where you can find out more about it.

WHAT IS BLUE RUBBER BLEB NAEVUS SYNDROME?

Blue rubber bleb naevus syndrome has also been called 'Bean syndrome' after Dr William Bean, who described its symptoms. It is caused by abnormal development of veins, known as venous malformations in the skin, soft tissues and gastrointestinal tract (gut or bowels).

A 'syndrome' is a group of signs and symptoms that occur together. 'Naevus' is Latin for 'birthmark'. The 'blue rubber blebs' are small growths made of blue mis-shaped veins with a rubber-like feel.

In some people, blebs are only seen in the skin. They can also occur internally, most commonly in the intestine.

WHAT CAUSES BLUE RUBBER BLEB NAEVUS SYNDROME?

The cause of blue rubber bleb naevus syndrome is not known. During the development of an affected baby in the womb, clusters of abnormal veins are formed. This is not known to be related to any event during pregnancy.

IS BLUE RUBBER BLEB NAEVUS HEREDITARY?

Although there are a few reports of blue rubber bleb naevus syndrome running in a family, this is not usually the case

Blue rubber bleb naevus syndrome is not contagious or cancerous.

WHAT DO BLUE RUBBER BLEBS FEEL AND LOOK LIKE?

Apart from their appearance, the skin lesions (growths) often do not cause any symptoms. However, they may be tender to touch, painful, show increased sweating or bleed. Sometimes, because of slow blood flow within lesions, the blood may clot, causing a warm, tender swelling.

Other symptoms depend on the involvement of other organs. This does not usually become apparent until adulthood. If the intestine is involved, blood may be passed when opening the bowels and the faeces may be black. Blood loss may result in anaemia. If you notice red or black stool, you should consult your GP.

Blue rubber blebs are groups of purple to bluish-black rubbery lumps that can be seen in the skin either at birth, or may appear in childhood or later in life. The number of blue rubber blebs usually increases during life and can range from a few to over a hundred. Pressing onto a bleb will cause it to blanche (go the colour of unaffected skin). On releasing the pressure, the blood will return, and the bleb will recover its previous colour.

HOW IS BLUE RUBBER BLEB NAEVUS SYNDROME DIAGNOSED?

A dermatologist may suspect blue rubber bleb naevus syndrome after examining the skin. It may be necessary to confirm the diagnosis by taking a sample of the abnormal skin (a biopsy) to be examined under the microscope. A local anaesthetic injection will usually be given beforehand to numb the area.

CAN BLUE RUBBER BLEB NAEVUS SYNDROME BE CURED?

Unfortunately, there is currently no cure for blue rubber bleb naevus syndrome.

HOW CAN BLUE RUBBER BLEB NAEVUS SYNDROME BE TREATED?

Some people require no treatment other than an explanation of the condition. Treatment is directed at the symptoms, which may vary from patient to patient and may be different depending on which parts of the body are affected.

For blebs of the skin, different surgical and laser treatments may be used, depending on regional availability. These require a local anaesthetic injection to numb the areas beforehand and can leave a scar:

- Excising (removing) lesions or scraping them off ('curettage') and treating the bleeding base with heat (cautery).
- Using a laser to remove the bleb layer by layer, such as the carbon dioxide (CO_2) laser, or the Erbium Yag laser.

Other treatment options include freezing the lesions with liquid nitrogen (cryotherapy) and sclerotherapy. Sclerotherapy is the injection of a solution into the veins of the bleb that causes inflammation and shrinks the veins.

Symptoms such as those related to anaemia, blood passed when opening bowels or black stool need to be investigated as they arise. In such cases, referrals to different specialists may be required.

There have reports that a medicine called sirolimus which dampens down the immune system might be helpful. It can potentially reduce the number or size of lesions on the skin and may reduce bleeding from the gut, or need for blood transfusions in people with gut lesions. Side effects of sirolimus, such as increased infections (due to its effect on the immune system) and inflammation of mucus membranes (like those in the eyes and mouth) can be reduced by giving a smaller dose.

Skin camouflage products can be useful to skin-match the bluish discolouration. They are water resistant and matched to the colour of the person's natural skin tone. More information and help are available from the charity Changing Faces.

WHERE CAN I GET MORE INFORMATION ABOUT BLUE RUBBER BLEB NAEVUS SYNDROME?

Patient support groups providing information:

Changing Faces

Web: www.changingfaces.org.uk

Tel: 0300 012 0275

Email: info@changingfaces.org.uk

Web links to other relevant sources:

http://dermnetnz.org/vascular/venous-malformation.html

http://rarediseases.info.nih.gov/gard/5940/blue-rubber-bleb-nevus-syndrome/resources/1

Please note that the British Association of Dermatologists (BAD) provides web links to additional resources to help people access a range of information about their treatment or skin condition. The views expressed in these external resources may not be shared by the BAD or its members. The BAD has no control of and does not endorse the content of external links.

This leaflet aims to provide accurate information about the subject and is a consensus of the views held by representatives of the British Association of Dermatologists: individual patient circumstances may differ, which might alter both the advice and course of therapy given to you by your doctor.

This leaflet has been assessed for readability by the British Association of Dermatologists' Patient Information Lay Review Panel

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