



Blue rubber bleb naevus syndrome

This information sheet from Great Ormond Street Hospital (GOSH) provides information about the causes, symptoms and treatment of blue rubber bleb naevus syndrome and where to get help.

What is blue rubber bleb naevus syndrome?

Blue rubber bleb naevus syndrome is the name given to a condition characterised by blue marks on the skin and internal organs caused by abnormal veins. The marks on the skin do not usually cause serious problems, but the ones affecting the internal organs can bleed, leading to anaemia and sometimes to impairment of clotting, and other symptoms depending on the location of the bleed.

What causes blue rubber bleb naevus syndrome?

Blue rubber bleb naevus syndrome is very rare, usually arising sporadically (by chance) with no other family members affected. In most cases, it arises as a result of mutation (change) in the TEK gene.

What are the signs and symptoms of blue rubber bleb naevus syndrome?

The blue marks on the skin, which may be present from birth, are the most obvious feature of blue rubber bleb naevus syndrome. The marks are very variable, both in terms of size and shape. They may occur anywhere externally or internally and may continue to appear throughout life.

Bleeding from venous malformations, most commonly in the digestive tract can cause anaemia. Anaemia is a condition where the number of red blood cells or the amount of haemoglobin in red

blood cells is less than normal. Haemoglobin is the substance that makes blood red and its main purpose is to carry oxygen around the body. The symptoms of anaemia include tiredness, weakness and lack of energy. Bleeding from venous malformations at other sites may be evident as pain, swelling and disturbance of function, depending on the site of the bleed.

How is blue rubber bleb naevus syndrome diagnosed?

The blue marks on the skin are usually visible immediately after birth. However, as blue rubber bleb naevus syndrome is such a rare condition, there may be some delay in diagnosis. As a rule, a group of specialists work together (multidisciplinary team) to ensure that care and treatment is coordinated.

In addition to a physical examination, MRI and endoscopy may be required to locate venous malformations in organs in other than the skin. An endoscope (a thin, flexible tube with a bright light at the end) is passed through your child's mouth and down into their stomach. The doctor can then look down the tube and have a clear view of the lining of the foodpipe, stomach and duodenum and can see whether there are any problems.

Blood tests will be required to look at the levels of haemoglobin and clotting factors in the blood. Biopsies from affected tissue, usually skin biopsies, are needed to look for the genetic mutations causing the condition

How is blue rubber bleb naevus syndrome treated?

No treatment may be required for the blue marks on the skin unless they are causing problems due to their appearance or location. Treatment of venous malformations at other sites, including the intestine, will depend on the location, the problems arising and the severity of anaemia and clotting disturbance. Treatment with a medication called sirolimus may be required in severe cases. If bleeding from the blue marks in the digestive system is causing anaemia, regular blood transfusions may be needed to maintain haemoglobin levels. Some blue marks in the digestive system can be treated using endoscopic laser treatment or bowel resection.

Further information and support

- At Great Ormond Street Hospital (GOSH), contact the Birthmark Unit.
- The Birthmark Support Group offers support and advice to parents of children with all types of birthmark, including blue rubber bleb naevus syndrome. Telephone their helpline on 0845 045 4700 or visit their website at www.birthmarksupportgroup.org.uk
- Changing Faces is the support organisation for anyone affected by visible difference. Telephone their helpline on 0845 4500 276 or visit their website at www.changingfaces.org.uk. They also have a specific website for teenagers and young adults at www.iface.org.uk