



Blue rubber bleb naevus syndrome

This information sheet from Great Ormond Street Hospital (GOSH) explains 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 where blue marks develop on the skin and internal organs. These blue marks are venous malformations, which occur when the blood vessel wall does not hold its shape as it should so blood flow through the area is slowed down. The marks on the skin do not usually cause any serious problems, but the ones affecting the internal organs can bleed, sometimes leading to anaemia.

What causes blue rubber bleb naevus syndrome?

Blue rubber bleb naevus syndrome is a very rare condition – only around 200 cases have been reported in the medical literature to date. In most of these cases, the condition developed sporadically (by chance) and no other family members were affected. However, in a small number of people, there seemed to be a genetic component. Again, from the cases reported, doctors believe that it affects males and females equally, but that it affects more people from a white background. Research is currently being undertaken to identify the gene for blue rubber bleb naevus syndrome.



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

The main feature of blue rubber bleb naevus syndrome is the blue marks on the skin. These can be present from birth and are soft and rubbery. They can be depressed painlessly and sometimes 'refill' quickly. The marks are very variable, both in terms of size and shape, and continue to appear throughout life. They can occur anywhere on the body including on internal organs but if they are over a joint, this can be uncomfortable.

The more serious features arise from venous malformations within the digestive system. These tend to appear in early adulthood and can bleed. This bleeding is not usually sudden or in large quantities but small amounts of bleeding over a long period of time 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.

How is blue rubber bleb naevus syndrome diagnosed?

The blue marks on the skin are clearly visible immediately after birth. However, as blue rubber bleb naevus syndrome is such a rare condition, there may be some confusion over the diagnosis. A full physical examination by doctors at a specialist centre is recommended, alongside clinical photography to record the location and size of all blue marks. In some cases, a group of specialists work together (multidisciplinary team) to ensure that care and treatment is coordinated.

In addition to a physical examination, endoscopy may be used to locate blue marks affecting the digestive system. 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.

If anaemia is suspected, blood tests to look at the levels of haemoglobin and clotting factors in the blood will be suggested.



How is blue rubber bleb naevus syndrome treated?

No treatment is required for the blue marks on the skin unless they are causing problems due to their appearance or location. The choice of medical, surgical and laser treatments will be determined by the location of the lesions and the problems that arise from them. Various medicines – such as thalidomide and rapamycin – can be used to reduce the spread and increase in blue marks. Further research is needed to understand how these medicines work.

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 congenital haemangioma. 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

Notes

Compiled by the Birthmark Unit in collaboration with the Child and Family Information Group

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