



Bernard-Soulier syndrome

This factsheet is about a bleeding disorder that is related to problems with a blood-clotting factor called Bernard-Soulier syndrome. It is written to go with our **Rare bleeding disorders booklet**, where you will find much more information on living with one of these conditions.

What is Bernard-Soulier syndrome?

Bernard-Soulier syndrome is a very rare platelet function disorder. There are around 96 people with Bernard-Soulier syndrome in the UK.

It is a bleeding disorder caused by an abnormality found in your genes (part of the DNA that you inherit from your parents). These genes affect a group of linked proteins (called a receptor) and are normally found on the surface of platelets a type of blood cell. Because this receptor is missing or is not working properly, platelets do not stick to the injured blood vessel wall as normal and a normal blood clot doesn't form properly.

What causes factor Bernard-Soulier syndrome?

This is an inherited genetic disorder that affects both males and females. It is what is

known as recessive, meaning you inherit the gene defect from both parents (even though they themselves do not have the disease). It affects men and women equally and can be found in all ethnic groups.

If you carry one copy of the gene fault, you are known as a carrier. You can only pass the condition on to your children if your partner also carries the gene fault. You will not have the condition yourself, but if you have children that inherit the gene fault from you, they will also be carriers of the condition.

Our **Rare bleeding disorders booklet** has more information about how bleeding disorders are inherited.

Symptoms of factor Bernard-Soulier syndrome

Most people with Bernard Soulier syndrome have some bleeding. The commonest symptoms are:

- bleeding after surgery, dental care or circumcision
- bleeding from the umbilical cord stump at birth
- bleeding in the gut (gastrointestinal system)
- blood in your urine

For everyone affected by a genetic bleeding disorder

To find out more, visit haemophilia.org.uk or contact us on 020 7939 0780 or at info@haemophilia.org.uk

- easy bruising
- heavier than usual bleeding after childbirth
- heavy periods that last longer than normal
- nosebleeds or bleeding into the mouth

It is very important that you contact your doctor or haemophilia centre if you think you or your child are having a bleed. Some bleeds can cause long-term problems if they are not properly treated, such as bleeds into muscles or joints. There is more information in our **Rare bleeding disorders booklet** about how to spot the different types of bleeds.

If you have frequent bleeds, such as nosebleeds, you can become anaemic. This means you have a low haemoglobin level because of the frequent loss of blood. Common symptoms of anaemia are due to the reduced amount of oxygen in the body. These include tiredness, having little energy (lethargy), feeling faint and becoming easily breathless. The treatment of anaemia depends on what is causing it. emia depends on what is causing it.

Diagnosing Bernard-Soulier syndrome

Bernard-Soulier syndrome is diagnosed with specialised blood tests, which should be done at a haemophilia centre. A careful medical history will also be taken. They will look closely at:

- Whether they find the missing receptor needed to help your blood clot in the blood sample your supply (using a test called flow cytometry).
- Whether your platelets clump together normally when something called ristocetin is added to your blood (substance that helps platelets to clump together).
- How long it takes for a platelet plug

to form in your sample of blood (known as 'the closure time) and whether this takes longer than normal.

Doctors will also look for

- Platelets that appear larger than normal under a microscope
- fewer platelets than normal

Treatment for Bernard-Soulier syndrome

Most people with platelet function disorders only need treatment during surgery or dental work and after injury or accidents. When needed, Bernard-Soulier syndrome may be treated with:

- antifibrinolytic drugs
- fibrin sealants
- hormonal contraceptives (the 'Pill') (to control excessive menstrual bleeding)
- iron replacement (if necessary to treat anaemia caused by excessive or prolonged bleeding)
- platelet transfusions (only if bleeding is severe)
- recombinant factor VIIa

You should not use Non-Steroidal Anti-Inflammatory Drugs (NSAIDs such as ibuprofen) as this increases the risk of bleeding. Other methods of pain relief should be used instead. Speak to your doctor if you are unsure.

Caution is needed for injections as well – immunisations should be given subcutaneously (under the skin) rather than intramuscularly (into a muscle) to reduce the risk of a painful bruised swelling (haematoma) developing.

Coping with your condition

Finding out that you or your child has a bleeding disorder can be upsetting and bring on a range of different emotions. Of course, this will take time to accept. Finding out as much as you can about your condition can help you learn to cope with it.

How much your bleeding disorder affects your daily life will depend on how severe it is.

Do find out as much as you can about how to prevent bleeding and when it is likely to cause a problem. Our **Rare bleeding disorders booklet** has a lot of information about what to look out for and precautions you can take to keep yourself healthy.

There is information on:

- carrying medical information with you
- dental care
- how to spot the early signs of a bleed
- information for girls and women about problems with periods and pregnancy
- ways to make bleeding less likely.

A new diagnosis can feel scary or overwhelming but there's lots of great support available.