The Platelet Charity
Helping people with bleeding disorders

Bernard-Soulier syndrome

What is it?

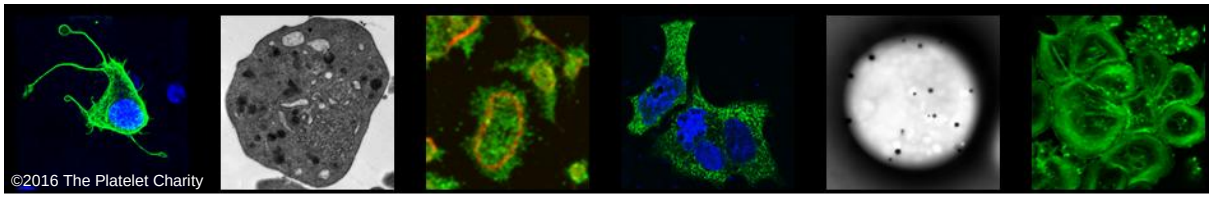
- Bernard-Soulier syndrome (BSS) is a well characterised and researched severe platelet bleeding disorder. It is usually detected at an early age, especially in families where other family members are affected.
- It was identified in 1948 by two French haematologists, Jean Bernard and Jean-Pierre Soulier
- There are several hundred patients worldwide, with approximately fifty patients in the UK.
- BSS is caused by a loss of one of three proteins on the platelet surface, GPIbα (also known as CD42), GPIbβ or GPIX. The three proteins, and a further protein, GPV, form a complex on the platelet surface known as the GPIb/IX/V receptor.
- All three proteins are only found on platelets and the platelet's mother cell, the megakaryocyte.
- The binding of a protein in the plasma, von Willebrand factor (VWF), to GPIb/IX/V captures platelets at sites of injury. In this context, VWF can be considered as a 'sticky glue'.

Who suffers?

- BSS affects males and females of all ages in approximately equal numbers. Two copies of the mutant gene are inherited (one each) from the mother and father (autosomal recessive). It is more common in families were there are relationships between individuals who are blood relatives of each other. In general carriers of the condition (who only have one mutated gene) do not experience any clinical problems, although there are exceptions.

What are the symptoms?

- Patients with BSS may experience nose bleeding, easy bruising, bleeding from gums, heavy or prolonged menstrual bleeding (menorrhagia), bleeding after childbirth, abnormal bleeding after surgery or dental work, and gastrointestinal bleeding.



Diagnosis

- Patients have a low platelet count (thrombocytopenia), enlarged platelets and a reduced platelet aggregation (clumping) response to ristocetin, which is a compound used when platelet function is investigated.
- Diagnosis is confirmed (by a technique called flow cytometry) using an antibody against the normal GPIb/IX/V receptor.
- A definitive diagnosis is made by genetic sequencing.

Treatment

- Treatment should be led by and discussed with a haematologist with experience in bleeding disorders.
- Treatment is the same as other conditions where there are platelet function defects. General strategies include avoidance of medications that inhibit platelet function further (e.g. aspirin) and compression at sites of injury. Patients may be treated with antifibrinolytics (e.g. tranexamic acid) or platelet transfusion prior to surgery.
- Patients should carry a bleeding disorders / haemorrhagic states card (issued by a Haemophilia Care Centre) to alert others to their condition.
- Genetic counselling may be offered.

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