Bernard-Soulier Syndrome

Bernard-Soulier syndrome is an autosomal recessive disorder, causing a deficiency of glycoprotein IB (GpIb) receptors on platelets. This inhibits platelet ability to agglutinate to vWF on damaged endothelial cells on vascular tissues. This manifests like other platelet disorders, causing mucous membrane bleeding, epistaxis, menorrhagia, and easy bruising. Labs show large platelets and increased bleeding time. Bernard-Soulier can be differentiated from Glanzmann's thrombasthenia because in this disorder, ristocetin cofactor assay does not lead to platelet agglutination.



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Mechanism

Autosomal Recessive
Recessive-chocolate
Bernard-Soulier syndrome is inherited in an autosomal recessive fashion.

Deficiency of Glycoprotein Ib (GpIb) Receptor
Broken Glider-protein with (1) Wand Bee
This inherited disorder leads to a deficiency of glycoprotein Ib (GpIb) receptors on platelets. Without these receptors, platelets cannot bind to vWF expressed by endothelial cells that are injured, which would normally lead to clotting at the site of vascular injury.

Symptoms

Mucosal Membrane Bleeding
Mucous Dripping and Bleeding
This disorder displays many common manifestations of platelet disorders. Patients are more prone to mucosal membrane bleeding, and may display recurrent epistaxis or bleeding gums. Furthermore, these patients often show prolonged postoperative bleeding.

Menorrhagia
Very bloody Tampon-with-rags
Menorrhagia is defined as menstruation on a regular cycle, but with excessive flow and duration lasting longer than seven days. Female patients with Bernard-Soulier syndrome may complain of this symptom.

Easy Bruising
Easy-button Bruising
As with other platelet disorders, patients show easy bruising with Bernard-Soulier syndrome.

Labs

Large Platelets
Large Plates
Bernard-Soulier syndrome is often referred to as a giant platelet disorder, as patients have large platelets on peripheral smear. The large platelets and low platelet count in BSS are seemingly due to the absence of GPIb and filamentous structures, which helps stabilize the platelet cytoskeleton. The platelet count can be decreased or unaffected.

Increased Bleeding Time
Up-arrow Blood Clock
With this disorder, a lack of normal platelet function leads to an increased bleeding time. It should be noted that the PT and PTT are unaffected in Bernard-Soulier syndrome.

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No Platelet Agglutination with Ristocetin

Plates not sticking with Red-stone-cement

A test that helps distinguish Bernard-Soulier syndrome from other platelet disorders is the ristocetin cofactor assay. In this disease, platelets do not aggregate to ristocetin, distinguishing it from Glanzmann thrombasthenia.

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