Bernard-Soulier Syndrome

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Risk

- Estimated to be <1 in 1 million persons, but may be higher due to misdiagnosis and underreporting
- Rare: Approximately 100 cases reported in literature

Perioperative Risks

- Severe hemorrhage out of proportion to plt count
- Transfusion reactions

Worry About

- Severe periop hemorrhage
- Limited availability of blood products
- Concurrent medical conditions (e.g., uremia, liver disease) or medications (NSAIDs, heparin, and antiplatelet agents) contributing to bleeding

Overview

- Coagulopathy characterized by defects in plt number and function due to an absence or abnormality in plt membrane glycoprotein receptor complex GPIb-IX-V, a four-protein complex responsible for initiating plt adhesion at sites of vascular injury and binding Von Willebrand factor
- Defect in primary hemostasis; mucocutaneous bleeding; often, the bleeding is more severe than expected for the pt's particular plt count
- Clinical phenotype severity varies; manifestations range from easy bruising, purpura, epistaxis, gingival bleeding, and menorrhagia to hematuria, GI bleeding, and fatal hemorrhage
- Severe bleeding associated with menses, trauma, and certain surgical procedures (e.g., tonsillectomy, appendectomy, splenectomy, dental extraction)
- Diagnosed by prolonged bleeding time, presence of a small number of very large plt on blood smears (macrothrombocytopenia), reduced plt counts (20,000–100,000), and absence of RIPA

Etiology

- Autosomal recessive inheritance pattern; a wide spectrum of clinical manifestation based on the degree of glycoprotein complex dysfunction.
- Individual genes have been identified for each of the proteins in the complex and may be the target for future therapy: 17p12 (GPIba), 22q11.2 (GPIbb), 3q29 (GPV), and 3q21 (GPIX).

Usual Treatment

- Bleeding prophylaxis including lifestyle modifications (e.g., personal safety, avoidance of trauma, avoidance of antiplatelet medications [aspirin], adequate dental hygiene, use of contraceptives in females at puberty)
- Bleeding treatment: Plts, PRBCs, and EACA
- Refractory bleeding: DDAVP, gamma globulin, corticosteroids, and recombinant factor VIIa

Assessment Points

System	Effect	Assessment by Hx	PE	Test
HEENT	Oral/mucosal friability	Epistaxis and gingival and cutaneous bleeding	Sores, stomatitis, erythema	See HEME
CV	Vascular access: potential hemorrhage			
GI	GI bleeding	Melena, hematochezia		Stool guaiac, endoscopy
HEME	Coagulopathy: Primary hemostasis, mucocutaneous bleeding Severe hemorrhage Antibodies to blood products	Bleeding gums, bruising easily, menorrhagia, epistaxis Hx of transfusion family Hx of periop bleeding	Petechiae, bruises, gingival hyperemia	PT/ INR, PTT, plt count, blood smear, plt function assay, ristocetin cofactor activity Type and screen, crossmatch, and antibody analysis

Key References: Kostopanagiotou G, Siafaka I, Sikiotis C, et al: Anesthetic and perioperative management of a patient with Bernard-Soulier syndrome, *J Clin Anesth* 16(6):458-460, 2004; Lanza F: Bernard-Soulier syndrome (hemorrhagiparous thrombocytic dystrophy), *Orphanet J Rare Dis* 1:46, 2006.

Perioperative Implications

Preoperative Preparation

- Collaboration with hematology and blood bank.
- Ensure availability and adequacy of blood products.
- Assess and optimize coagulation (coagulation factor analysis, dialysis if uremic, and FFP/vitamin K if increased INR).

Monitoring

- Standard monitors.
- Risk-benefit assessment to evaluate more access or invasive monitoring (A-line or CVP) versus unnecessary or failed attempts leading to sources of potential bleeding.
- Urine output for new-onset hemoglobinuria as first sign of transfusion reaction.
- Avoid undue tension on soft tissues and provide adequate padding of pressure points and mucosal surfaces.
- Consider intraop thromboelastogram.

Airway

- Avoid nasal manipulation.
- Use extreme caution with friable oral and pharyngeal mucosal surfaces.
- Consider video laryngoscopy to ensure first-attempt success.

Induction

- No specific recommendations

Maintenance

- Avoid hemodilution.
- Meticulous surgical hemostasis.
- Normothermia promotes coagulation.
- Analyze clot formation via thromboelastography and transfusion as needed.
- Controlled hypotension may reduce potential blood loss; however, avoid in anemic pts.

Extubation

- Care of mucosal membranes, gentle orotracheal suction under direct visual guidance, and avoid coughing

Adjuvants

- Neuraxial anesthesia is relatively contraindicated in these pts. Individual risk-benefit assessment based on severity of disease and plt function (e.g., thromboelastography).

Postoperative Period

- Continue monitoring coagulation status.

Anticipated Problems/Concerns

- Severe intraop and postop hemorrhage
- Transfusion-related reaction and increased likelihood of infectious bloodborne diseases

Bilirubinemia of the Newborn

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Risk

- A common problem in neonates.
- Some types pathologic and some physiologic.
- Bilirubin may be unconjugated or conjugated; differentiating important for diagnosis.
- If pathologic, varying effect on management (e.g., sepsis, Rh incompatibility, GI obstruction, Gilbert, AVM, sickle cell, biliary atresia).
- Clinical, epidemiologic, and genetic risk factors associated with significant hyperbilirubinemia include preterm gestational age, exclusive breastfeeding, glucose-6-phosphate dehydrogenase deficiency, Rh/ABO incompatibility, East Asian or Native American ethnicity, any jaundice observed in the first 24 h of life (hemolysis until proven otherwise), cephalohematoma or significant bruising after delivery, and Hx of a previous sibling treated with phototherapy.

Perioperative Risks

- Risks specific to a primary pathologic cause of bilirubinemia
- Acute bilirubin encephalopathy (unconjugated bilirubin may penetrate brain cells and cause dysfunction in either pathologic or physiologic states)
- Kernicterus (chronic and permanent sequelae of bilirubin neurotoxicity)

Worry About

- Factors that increase blood-brain barrier permeability to unconjugated bilirubin (hypoxia, hypercarbia, acidosis, hyperosmolality, hypertension, seizure activity, and sepsis)
- Drugs (e.g., sulfonamides, ceftriaxone, ampicillin, salicylates, furosemide, contrast dye) that displace bilirubin from albumin, which can increase free fraction of unconjugated bilirubin in the blood