NOTES
PLATELET DISORDERS

GENERALLY, WHAT ARE THEY?

PATHOLOGY & CAUSES

- Platelet dysfunction, impaired hemostasis, bleeding

RISK FACTORS

- Children of parents who are close relatives (consanguineous)

COMPLICATIONS

- Mild to severe hemorrhage

SIGNS & SYMPTOMS

- Mucocutaneous bleeding
- Excessive bruising/bleeding after minor trauma
- Immediate, excessive bleeding with invasive procedures

DIAGNOSIS

LAB RESULTS

- Complete blood count (CBC)
- Clotting studies (e.g. bleeding time)
- Platelet function tests

TREATMENT

- Manage spontaneous/trauma-related bleeding episodes

MEDICATIONS

- Anti-fibrinolytic therapy
- Platelet transfusions
- Avoid antiplatelet medications
- Corticosteroids, immunosuppressants

BERNARD–SOULIER SYNDROME (BSS)

osms.it/bernard-soulier-syndrome

PATHOLOGY & CAUSES

- Rare, inherited clotting disorder; mild thrombocytopenia, macrothrombocytopenia (giant platelets), platelet dysfunction, bleeding

CAUSES

- Autosomal recessive inheritance pattern
- Platelets lack essential glycoprotein Ib-IX-V complex (GPIb) → impaired hemostasis → bleeding

RISK FACTORS

- Prevalence in individuals of Mediterranean descent
- Children of parents who are close relatives (consanguineous)

SIGNS & SYMPTOMS

- Mucocutaneous bleeding
  - Epistaxis; gingival bleeding; petechiae, purpura (coalesced petechiae); GI bleeding; genitourinary bleeding (e.g. hematuria); menorrhagia
- Excessive bruising/bleeding after minor trauma; bruises linger
- Immediate, excessive bleeding with invasive procedures
- Asymptomatic until adulthood

DIAGNOSIS

LAB RESULTS

- CBC
  - Low platelet count
- Peripheral blood smear analysis
  - Giant platelets (accelerated platelet turnover)
- Clotting studies
  - Bleeding time prolonged (PT, aPTT normal)
- Flow cytometry
  - Deficient/absent GPIb-IX-V complex

OTHER DIAGNOSTICS

- Physical examination
  - Purpura
  - Ecchymoses

TREATMENT

MEDICATIONS

- Avoid antiplatelet medications
- Anti-fibrinolytic therapy (e.g. tranexamic acid)

OTHER INTERVENTIONS

- Platelet transfusions (e.g. prophylaxis before invasive procedures)
  - HLA matching/leukocyte reduced platelets reduces risk of allo-antibody formation

GLANZMANN'S THROMBASTHENIA (GT)

osms.it/glanzmanns-thrombasthenia

PATHOLOGY & CAUSES

- Inherited bleeding disorder, defect in platelet surface receptor $\alpha_{IIb}\beta_3$
- Platelet-mediated hemostasis
  - Binding of platelets to exposed components of injured endothelium through glycoprotein (GP) receptors on platelet surface (e.g. GPIb/IX, GPIa/IIa, integrin $\alpha_{IIb}\beta_3$)
- Integrin $\alpha_{IIb}\beta_3$ defect → impaired platelet clot retraction, altered hemostasis

CAUSES

- Autosomal recessive inheritance pattern
- Rarely, allo-/auto-antibodies to platelet $\alpha_{IIb}\beta_3$ produced by autoimmune conditions (e.g. systemic lupus erythematosus, immune thrombocytopenia, myelodysplastic syndrome) during pregnancy/with use of platelet integrin $\alpha_{IIb}\beta_3$ antagonists (abciximab, eptifibatide)

RISK FACTORS

- Slightly more common in individuals who are biologically female
- Children of parents who are close relatives

- Conditions requiring frequent platelet transfusion
  - Platelet alloimmunization

COMPLICATIONS

- Fatal bleeding
  - Risk increases during childbirth (maternofetal bleeding, primary/secondary postpartum hemorrhage)

SIGNS & SYMPTOMS

- Mucocutaneous bleeding
  - Epistaxis; gingival bleeding; petechiae, purpura (coalesced petechiae); gastrointestinal (GI) bleeding; genitourinary bleeding (e.g. hematuria); menorrhagia
- Excessive bruising/bleeding after minor trauma
- Immediate, excessive bleeding with invasive procedures
- Infants
  - Leukocytosis, delayed separation of umbilical cord, purpura, spontaneous bruising, mucocutaneous bleeding

DIAGNOSIS

LAB RESULTS

- CBC
  - Platelet count normal
- Clotting studies
  - Bleeding time prolonged (PT, aPTT normal)
- Light transmission aggregometry (LTA)
  - Determines degree of platelet aggregation
  - Decreased or absent in GT
- Platelet function analyzer (PFA)
  - Measures flow rate as platelets form platelet plug within capillary tube
  - Formation of platelet plug prolonged in GT
- Flow cytometry
  - Deficient/absent $\alpha_{IIb}\beta_3$ platelet receptors
  - Mutation analysis through genomic DNA sequencing

OTHER DIAGNOSTICS

- Physical examination
  - Purpura
  - Ecchymoses

TREATMENT

- Avoid antiplatelet medications, punctures, invasive procedures

MEDICATIONS

- Individuals who are biologically female, of childbearing age
  - *Metrorrhagia:* oral contraceptives (suppress menstrual periods); iron supplementation; hysterectomy (if bleeding severe)
  - *Childbirth:* prophylaxis with recombinant factor VIIa + antifibrinolytic agent
- Rituximab, corticosteroids, immunosuppressants (e.g. cyclophosphamide)

OTHER INTERVENTIONS

- Manage bleeding episodes
  - Compression, fibrin sealants, gelatin sponges, nasal packing, topical thrombin, anti-fibrinolytic therapy (e.g. tranexamic acid), recombinant factor VIIa
- Platelet transfusions (e.g. prophylaxis before invasive procedures)
  - HLA matching/administration of leukocyte reduced platelets reduces allo-antibody formation
- Oral hygiene mitigates gingival bleeding
- Hematopoietic cell transplantation (if bleeding severe, recurrent)