Blood Dyscrasias

Noun – condition in which constituents of blood are abnormal or present in abnormal amounts.

Conditions related to hemostasis:

Coagulation-promoting conditions

- Procoagulant afibrinogenemia/dysfibrinogenemia
- Protein C deficiency (essential Vitamin K-dependent coagulation protein)
- Protein S deficiency (essential Vitamin K-dependent coagulation protein)
- Antithrombin III deficiency (essential coagulation protein, inhibits thrombin)
- Factor V Leiden deficiency
- Activated protein C resistance (aPCr) (inactivates factors Va and Villa, halting coagulation)
- Disseminated Intravascular Coagulopathy (widespread clotting within vessels)

Coagulation-impeding conditions

- Anticoagulant afibrinogenemia/dysfibrinogenemia
- Factor V deficiency (Autosomal recessive deficiency)
- Factor VII deficiency (Autosomal recessive or acquired in association with vitamin K deficiency, factor VII is a vitamin K-dependent protein required in the extrinsic coagulation pathway).
- Factor X deficiency (Autosomal recessive or acquired leading to reduced activity of both intrinsic and extrinsic pathways, etc.)
- Factor XI deficiency (Autosomal recessive, it’s a key component of the intrinsic pathway)
- Factor XII deficiency (Reduced or absent Hageman factor which initiates the intrinsic pathway. A deficiency makes monitoring heparin-induced anticoagulation useless).
- Factor XIII deficiency (Autosomal recessive or acquired, it stabilizes fibrin, preventing clot formation).
- Hemophilia A (missing factor VIII)
- Hemophilia B (missing factor IX)
- Hypoprothrombinemias (missing Prothrombin)
- Thrombocytopenias (reduced number of platelets)

Nonplatelet vascular and nonvascular hemostatic disorders

Abnormal circulating protein-related disorders

- Cryoglobulinemias (immunoglobulins that react to cold temperatures)
- Multiple myeloma (see WBCs)
- Waldenstrom macroglobulinemia (excessive IgM production)

Purpuras (bruising, unusual bleeding)

- Henoch-Schonlein purpura (hypersensitivity vasculitis seen in kids)
- Hyperglobulinemic purpura (too much gamma immunoglobulin)
- Waterhouse-Friderichsen syndrome (caused by septicemia)
- Wiskott-Aldrich syndrome (rare X-lined immunodeficiency seen only in young boys)
- Thrombotic thrombocytopenic purpura (familial and acquired forms – reduction in platelets due to the formation of abnormal clots)

Other

- Cavernous hemangioma (vascular tumors composed of large, dilated blood vessels)
- Hereditary hemorrhagic telangiectasia (autosomal dominant vascular malformations)
- Pseudoxanthoma elasticum (genetic collagen disorder)
- Ehlers-Danlos and Marfan syndrome (genetic collagen disorders)
- Scurvy (connective tissue disorder due to lack of vitamin C)
- Shwartzman phenomenon (Systemic vasculitis that starts with endotoxin exposure)
- Vitamin K deficiency
- Hemorrhagic disease of newborns (hereditary hemorrhagic telangiectasia – blood vessels are malformed and weak, thus prone to bleeding)
- Von Willebrand disease (Autosomal recessive deficiency or absence of vWF factor which acts to adhere platelets to each other in a clot)
- Antiphospholipid Antibody syndrome (autoimmune disorder in which antibodies are produced to the phospholipids in cell membranes, including in the cell membranes of endothelial cells which causes clots to form)

Other cell-related diseases
Blood Dyscrasias

RBCs and platelets (thrombocytes)
- Anemia (too few RBCs)
  - Aplastic anemia (bone marrow isn’t producing RBCs, WBCs or platelets)
  - Chronic disease-related anemia (especially cancer, inflammation or infections)
  - Excessive bleeding
  - Fanconi anemia ( Autosomal recessive, rare form of aplastic anemia)
  - Hemolytic anemia (anemia caused by destruction of RBCs)
    - Glucose-6-Phosphate Dehydrogenase Deficiency
    - Autoimmune
  - Iron deficiency anemia (not enough hemoglobin)
  - Pernicious or megaloblastic anemia (lack of Vitamin B12 or folate which are required to make RBCs)
  - Sickle cell anemia (abnormal hemoglobin causes RBCs to sickle)
  - Thalassemia (Autosomal recessive abnormal hemoglobin leads to a type of anemia)
- Hemochromatosis ( Autosomal recessive or acquired, leads to too much iron, which then builds up in organs)
- Idiopathic thrombocytopenic purpura (immune thrombocytopenic purpura – autoimmune condition where antibodies are produced to platelets, may be drug-induced).
- Thrombocytopenia (unknown cause of too many platelet-precursor cells, megakaryocytes)
- Thrombocytosis (known cause of too many platelets – another disease or condition)
- Thrombocytopenia (too few platelets)

WBCs
- Lymphocytopenia or lymphopenia (too few lymphocytes, B lymphocytes, T lymphocytes and natural killer cells)
- Lymphocytosis or lymphocytic leukocytosis (too many lymphocytes)
  - Cancers of lymphocytes (B, T or natural killer cells) or their precursors
    - Acute lymphocytic leukemia (lymphocyte precursor becomes cancerous)
    - Chronic lymphocytic leukemia
      - Usually B cells (e.g., Hairy cell leukemia)
      - Sezary syndrome (T-cell leukemia)
    - Lymphoma (cancers of mature B or T cells)
      - Hodgkin Lymphoma (B or T cell)
      - Non-Hodgkin lymphoma (B or T cell) – more than 20 different diseases
        - Burkitt’s lymphoma (B cell)
        - Mycosis fungoides (T cell)
- Plasma cell (B lymphocytes) disorders
  - Monoclonal gammopathies (one noncancerous B cell clone multiplies uncontrollably – about 25% of cases progress to cancer)
  - Multiple myeloma (cancer of plasma cells)
  - Macroglobulinemia (cancer in which a single B cell clone produces excess IgM, or macroglobulins)
  - Heavy chain diseases
    - Alpha heavy chain disease (cancerous B cells invade GI mucosa causing severe diarrhea and weight loss)
    - Gamma heavy chain disease (cancerous B cells invade bone marrow – may be asymptomatic)
    - Mu heavy chain disease (cancerous B cells invade liver, spleen and lymph nodes)
- Cancers of the precursors of neutrophils, basophils, eosinophils and monocytes
  - Acute myelocytic leukemia
    - Acute promyelocytic leukemia (precursors of neutrophils have a genetic mutation that prevents the binding and activity of Vitamin A which then prevents the normal maturation into neutrophils)
  - Chronic myelocytic leukemia
- Neutropenia (too few neutrophils)
- Neutrophilic leukocytosis (too many neutrophils)
- Eosinophilia (too many eosinophils)
- Idiopathic Hypereosinophilic syndrome (too many eosinophils, usually without obvious cause)
- Monocytosis (too many monocytes, the precursors of macrophages)
- Monocytopenia (too few monocytes)
- Basopenia (too few basophils)
- Basophilia (too many basophils – hyperthyroidism and myeloproliferative disorders like polycythemia vera are causes)

Bone marrow
- Myelofibrosis (fibrocytes replace blood-produceing cells in bone marrow and lay down excess fibrous tissue resulting in abnormal RBCs, anemia and an enlarged spleen)
- Polycythemia vera (bone marrow produces too many RBCs, usually due to a gene mutation, but may be acquired)