

Thalassemia

Autosomal recessive genetic disorder of inadequate production of normal hemoglobin. found in mediterranean ethnic groups.

Clinical Manifestation

Asymptomatic | Major retardation | Life threatening

Splenomegaly, Heptomegaly

Pathophysiology

Genetic defect, synthesis of one of a or b globin chains, production of abnormal Hgb and RBC, Hemolysis, Anemia

Collaborative Care

- No specific drug or diet are effective for treatment
- Minor (hetero-beta) : body adapts to low Hgb
- Major (Homo-beta) : Blood transfusions with IV deferoxamine (used to remove excess iron from the body)

Polycythemia

A net increase in the total number of Red blood cells. **Overproduction may be due to:**

- myeloproliferative syndrome in bone marrow
- reaction to chronically low oxy lvls or malignancy

Pathophysiology

Myeloproliferative due to hypoxia, the stem cell grow uncontrollably, BM becomes Hypercellular, Blood becomes thick to sluggish circulation, BM becomes fibrotic.

Complications

- Increased viscosity of blood
- Hemorrhage and thrombosis

Treatment

- Phlebotomy
- Myelosuppressive agents : Intron A, Agrylin, Aspirin

Idiopathic Thrombocytopenia Purpura (ITP)

Disorder of decreased platelets (below 150,000)

Causes

- Low production of platelets
- Increased breakdown of platelets

Symptoms

- Bruising, Nosebleeds, Petechiae

Immune Thrombocytopenic Purpura

- Abnormal destruction of circulating platelets
- Autoimmune disorder
- Destroyed in hosts' spleen by macrophages

Thrombotic Thrombocytopenic Purpura

- Increase agglutination of platelets, form microthrombi

Diagnostic Studies

- Platelet count, Hgb/Hct, prothrombin time (PT)
- Activated partial thromboplastin time (aPTT)

Treatments

- Corticosteroids (Bethamethasone, prednisone, predlone)
- Plasmapheresis, Splenectomy, platelet transfusion



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Page 1 of 1.

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