HEREDITARY/ACQUIRED HEMOLYTIC ANEMIA

HEMOLYTIC ANEMIAS

Hemolytic anemias = reduced red-cell life span

Classification of Hemolytic anemias

I. Red cell abnormality (Intracorpuscular factors)

A. Hereditary

1. Membrane defect (spherocytosis, elliptocytosis)
2. Metabolic defect (Glucoze-6-Phosphate-Dehydrogenaze (G6PD) deficiency, Pyruvate kinase (PK) deficiency)
3. Hemoglobinopathies (unstable hemoglobins, thalassemias, sickle cell anemia)

B. Acquired

1. Membrane abnormality-paroxysmal nocturnal hemoglobinuria (PNH)

II. Extracorpuscular factors

A. Immune hemolytic anemias

1. Autoimmune hemolytic anemia
   - caused by warm-reactive antibodies
   - caused by cold-reactive antibodies
2. Transfusion of incompatible blood

B. Nonimmune hemolytic anemias

1. Chemicals
2. Bacterial infections, parasitic infections (malaria), venons
3. Hemolysis due to physical trauma
   - hemolytic - uremic syndrome (HUS)
   - thrombotic thrombocytopenic purpura (TTP)
   - prosthetic heart valves
4. Hypersplenism

SOME TYPES OF HHA eg.

- SICKLE CELL DISEASE
- THALASSEMIAS
- G6PD DEFICIENCY
- HEREDITARY SPHEROCYTOSIS

G6PD DEFICIENCY

- MOST COMMON HUMAN ENZYME DEFECT
- X-LINKED DISORDER
- AFFECTS 15% OF U.S. BLACK MALES
- DECREASE IN GLUTATHIONE LEVELS

G6PD DEFICIENCY

- HEINZ BODIES SEEN ON PERIPHERAL BLOOD SMEAR
- NEONATAL JAUNDICE 1-4 DAYS AFTER BIRTH IN SEVERE VARIANTS
- INCREASE INCIDENCE OF PIDMENTED GALLSTONES AND SPLENOMEGALY

G6PD DEFICIENCY

- ACUTE HEMOLYTIC CRISIS DUE TO:
  - BACTERIAL/VIRAL INFECTION
  - OXIDANT DRUGS (*SULFAMETHOXAZOLE*)
  - METABOLIC ACIDOSIS (DKA)
  - RENAL FAILURE
  - INGESTION OF FAVA BEANS

G6PD DEFICIENCY

- DIAGNOSIS – QUANTITATIVE ASSAY DETECTING LOW ENZYME
- TREATMENT – SUPPORTIVE AND PREVENTATIVE

HEREDITARY SPHEROCYTOSIS

- RBS MEMBRANE DEFECT
- MOST COMMON HEREDITARY ANEMIA FROM PTS OF NORTHERN EUROPEAN DESCENT
- AUTOSOMAL DOMINANT
- MUTATIONS IN SPECTRIN AND ANKYRIN (MEMBRANE PROTEINS)

HEREDITARY SPHEROCYTOSIS

- **SPHEROCYTES** – IN PERIPHERAL BLOOD SMEAR
- SPHEROCYTES UNABLE TO PASS THROUGH THE SPLEEN
- SEVERE CASES REQUIRE A SPLENECTOMY

HEREDITARY SPHEROCYTOSIS

- NEONATAL JAUNDICE IN 1ST WEEK OCCURS IN 30-50% OF HS PTS
- ANEMIA, SPLENOMEGALY, JAUNDICE, AND TRANSFUSIONS NEEDED VARY DEPENDING ON SEVERITY OF DZ

Hereditary microspherocytosis

1. Pathophysiology
   - red cell membrane protein defects (spectrin deficiency) resulting cytoskeleton instability
2. Familly history
3. Clinical features
   - splenomegaly
4. Laboratory features
   - hemolytic anemia
   - blood smear-microspherocytes
   - abnormal osmotic fragility test
   - positive autohemolysis test
   - prevention of increased autohemolysis by including glucose in incubation medium
5. Treatment
   - splenectomy

Acquired Hemolytic Anemia

Introduction

- Increased RBC Destruction –
- Short RBC life span <120 days.
- Normocytic normochromic, reticulocytosis
- Anemia, Jaundice, marrow hyperplasia
- Splenomegaly, bilirubin gall stones
- Unconjugated “acholuric” (pale urine)
- Common types - AIHA, MAHA

Types of acquired HA

- AutoImmune Haemolytic Anemias (+ve DAT)
- Alloimmune haemolytic anemias
- Drug-induced immune haemolytic anemias
- Red cell fragmentation syndromes
- Infections
- Chemical & physical agents
- Secondary Haemolytic anemias
- Paroxysmal Nocturnal Haemoglobinuria (PNH)