HEREDITARY SPHEROCYTOSIS:

Def: Rare hemolytic Anemia Resulting from inherited defect in Red cell membrane. Lead to formation of spherocytes. These RBC are highly susceptible to sequestration & destruction in the spleen.

PATHOGENESIS:

Normal structure of RBC membrane. consist of, protein band 3 & glycophorin A via linker proteins namely Ankyrin & band 4.1

- usually a Autosomal dominant.
- cause due to mutation involve band 3, protein 4.2, Ankyrin or spectrin
- spherical RBC → due As a Result of destruction of RBC lipid membrane (destabilize). Young RBC Remain same shape, As they age RBC shed into circulation. — small Amount of cytoplasm is lost. As a Result they become spherical.
- Extravascular hemolysis: spherocyte are Rigid as a Result they get trapped in spleen → lead to destruction of RBC.

→ splenectomy is beneficial H-S.

Lab Findings: Hb↓, MCV↓, MCHC↑, Reticulocyte count↑

Peripheral Blood.

→ RBCs → Spherocytes, are small, dark, (hyperchromic) RBC without Any central Pallor.

Reticulocytosis → extensive RBC destruction. lead to compensatory hyperplasia of RBC progenitor in marrow.

WBC → TLC Raised

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- Bone Marrow:
  - Cellularity : Markedly hyper cellular
  - M:E Ratio : ↑ (8:1)
  - erythropoiesis : Erymoid hyperplasia
  - Megakaryopoiesis : Normal.
- Autohemolysis Test:
  - ↳ shows marked ↑ in spontaneous Autohemolysis.
- Direct coombs Test: +ve

BIOCHEMICAL FINDING:

Raised serum bilirubin

↑ Urine urobilinogen.

↓ haptoglobin

osmotic fragility ↑.

CLINICAL FEATURES: Age Anytime from neonatal to Adulthood

Family hait

→ Anemia : Mild to Moderate

→ Jaundice : Intermittent Attacks

→ splenomegaly

→ Gall stones - Pigmented occurs in 50-60%

Cause of spherocytosis is HS

→ Burns

→ Hypersplenism

→ Hemolytic transfusion

→ ABO drugs.