Erythrocytes Membrane Defects

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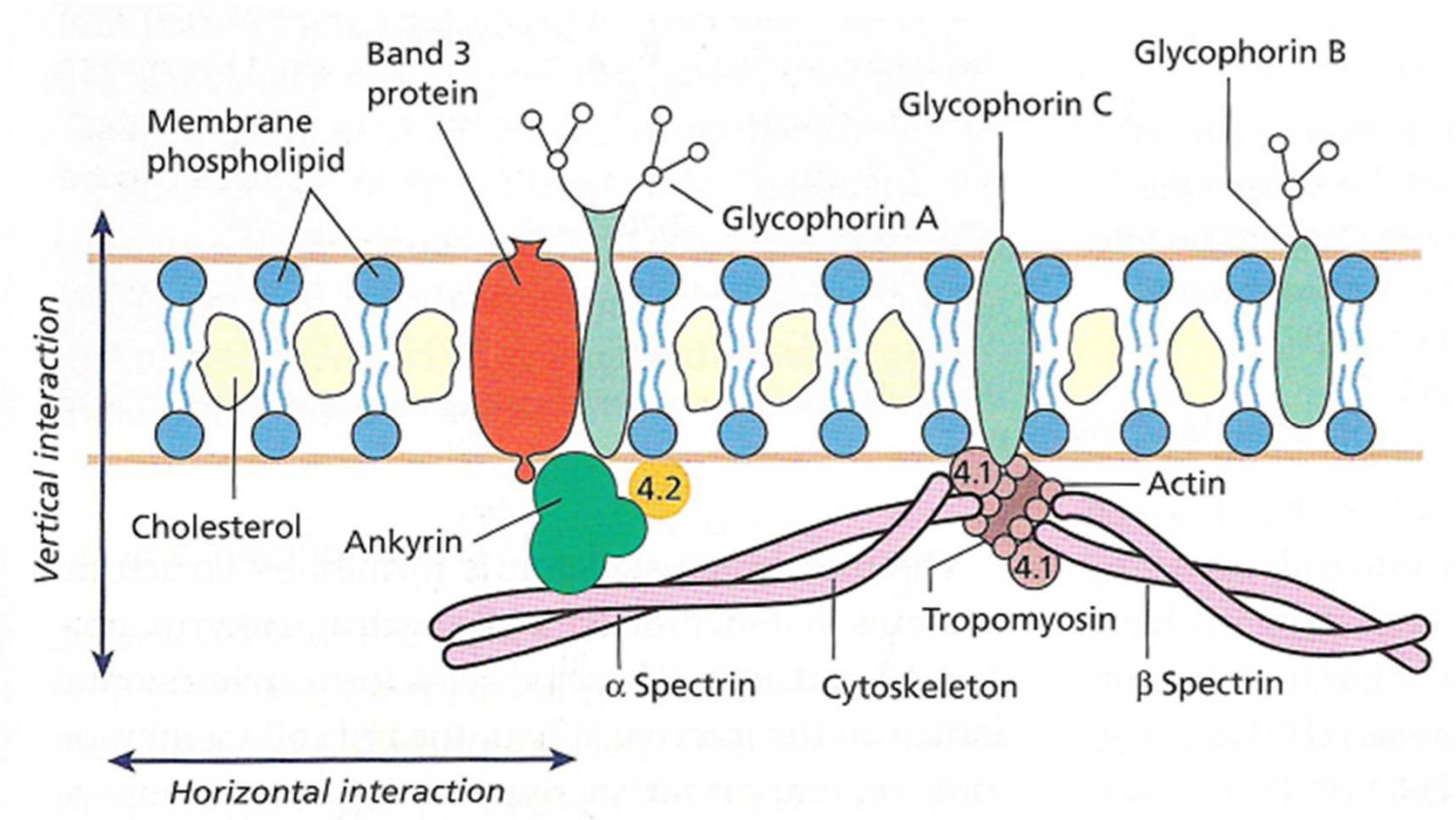
The Red Cell Membrane

- The red cell membrane consists of three main components:
- 1-phospholipid bilayer.
- 2- Various integral membrane proteins and.
- 3- Glycoproteins embedded in the phospholipid bilayer.

The red cell membrane structure



Red Cell Membrane Defects

- Abnormalities in the red cell membrane can result in hemolytic anemia of variable severity.
- The most common inherited membrane defects are:
  - hereditary spherocytosis and …
  - hereditary elliptocytosis

Hereditary Spherocytosis

Epidemiology:-

- **Hereditary spherocytosis (HS) is the most common inherited hemolytic anemia in people of Northern European descent.**
- **It also occurs in Asia and Africa**

Pathophysiology

- The fundamental cause in most cases of HS is defective vertical attachment between the phospholipid bilayer and the cytoskeleton.
- This results in loss of phospholipids from the cell membrane.
- Consequently, the surface area of the RBC decreases, and the cell gradually assumes the shape of a sphere.
- Spherocytes are less flexible than the normal RBCs, and tare therefore destroyed in the spleen.

Clinical Manifestations

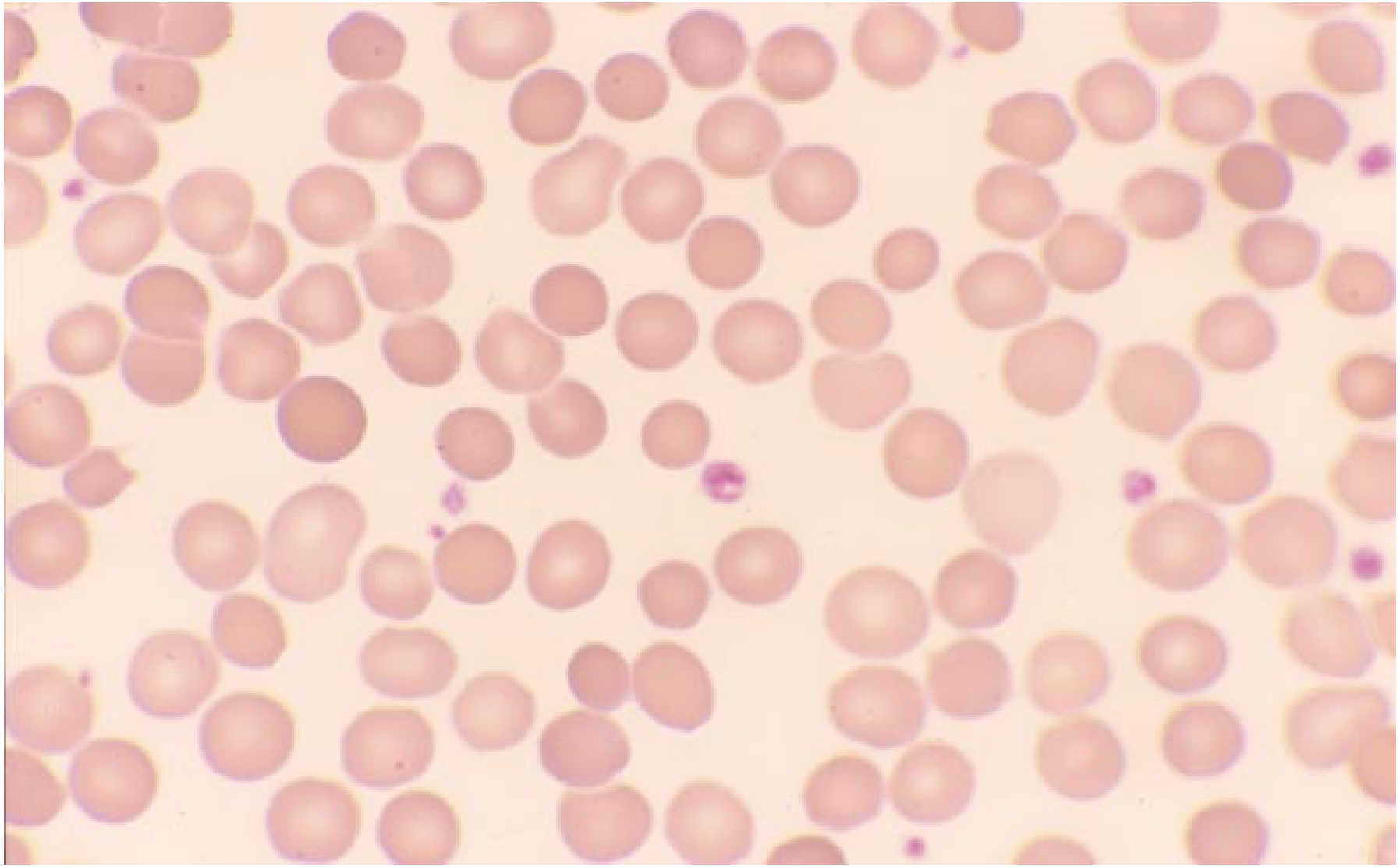
- The clinical manifestations of HS vary from asymptomatic to severe chronic hemolysis.
- Neonatal hyperbilirubinemia is frequent and may require exchange transfusion.
- The majority of older patients have relatively mild or moderate anemia, and the primary manifestations are hyperbilirubinemia and mild splenomegaly.

Lab. Diagnosis of HS

CBC:

- **MCV: Normal to slightly low.**
- **MCHC: Increased**
- **PBP: Microspherocytes**
- **Reticulocytes: Increased (~5–20%).**
- **Direct antiglobulin (Coombs') test: Negative.**

Blood smear shows spherocytosis



Osmotic fragility test:

- It is the classic laboratory test for HS.
- In this test, erythrocytes are incubated in saline solutions with osmolality ranging from normal to pure water.
- Percent hemolysis is measured by spectrophotometry.
- Erythrocytes from patients with HS hemolyze at higher saline concentrations than normal cells.

Hereditary Elliptocytosis

Epidemiology:

- **Most cases of hereditary elliptocytosis (HE) are inherited in an autosomal dominant pattern.**
- **It is found in Malaysia, New Guinea, Indonesia, and the Philippines.**
- **The geographic distribution of HE and its variants suggests a relationship to malaria (malaria *again*!).**

Pathophysiology

- Hereditary elliptocytosis is due to defective horizontal stability in the cytoskeleton.
- Red blood cells are squeezed into an elliptical shape as they pass through capillaries, and eventually RBCs become fixed in that shape.

Clinical Manifestations

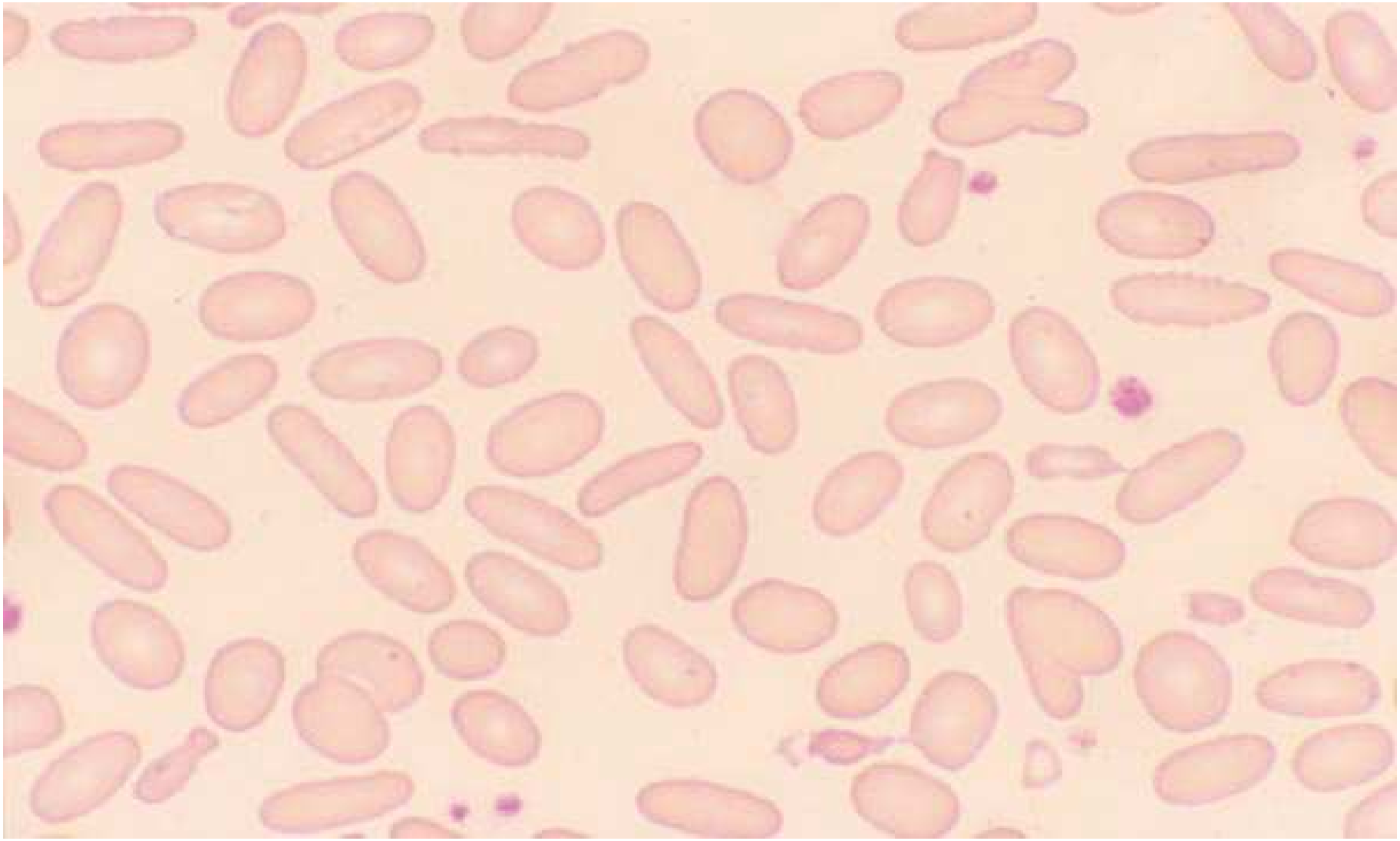
- Most cases of HE are asymptomatic, with normal RBC survival and no anemia.
- Neonatal hyperbilirubinemia is common and may require exchange transfusion.
- Some infants with HE have severe hemolytic anemia at birth, with marked microcytosis and poikilocytosis.

Diagnosis of Hereditary Elliptocytosis

PBP:

- It is the key diagnostic test.
- The presence of more than 15% elliptocytes suggests HE.
- Examining blood smears from parents or siblings may help confirm the presence of an inherited abnormality.
- The osmotic fragility test is normal.

Blood Smear Shows Elleptocytosis



THANK YOU