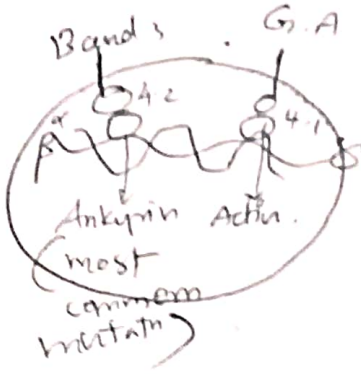
Hereditary Spherocytosis :-

↳ Inherited Autosomal Dominant.



- Most common defect in hereditary Spherocytosis is in Ankyrin

⇒ Most common defect in hereditary elliptocytosis is in spectrine

Pathogenesis

Mutation

↓

Loss of membrane cytoskeleton proteins

↓

Reduced membrane stability

↓

Loss of membrane relative to cytoplasm during exposure to shear stresses.

↓

Cells becomes microspherocytes

↓

Trapped in to spleen (due to reduced deformability)

↓

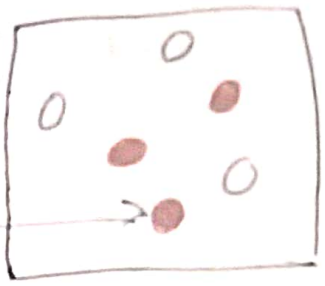
RBC are phagocytosed by RE cells.

↓

Extravascular Hemolysis.

Morphology of RBC

- Blood film shows the characteristic abnormality of erythrocytes in the form of microspherocytes



↓

fragile.

Special test – osmotic fragility test / Pink test.

Sickle Cell Anemia

Hemoglobinopathies.

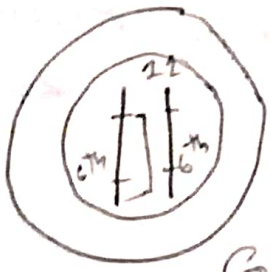
Qualitative disorders.

eg: Sickle Cell

Quantitative.

eg. Thalassemia.

β-chain



point mutation

6th Codon ~~chromosome~~ on chromosom 11

GAG → GTG

Glutamic acid → Valine.

HbA

HbS

↓

In deoxygenated state.

↓

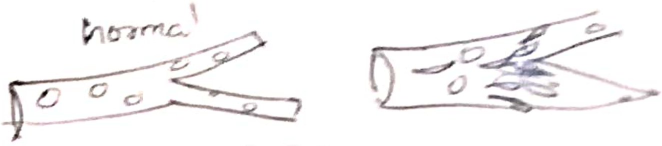
Sickle RBCs are sticky → causes Occlusion

↓

Ischemia of organs.

Consequences of Sickling

1. Vasoocclusion of microcirculation
2. Hemolytic anemia.
3. Increased MCHC.



C/F

1. Anemia
2. Vaso-occlusive phenomenon
   - → Bones – Hand-foot Syndrome
   - → Vertebra (fish mouth vertebra)
   - → Prominent cheek bones.
   - → Crew cut appearance (head on hair)

Spleen

- Initial (?ses) (splenomegaly)
- Later (1ses Rce) (Autosplenectomy – due to vasoocclusion by the sickled cells the spleen become atrophied due to Ischemia)