Haemoglobinopathies (Sickle Cell anemia)

- Autosomal recessive disorder when glutamic acid is substituted by valine at 6th position

Homologous
↓
HbS (Sickle cell disease)

Heterologous
↓
HbAS HbS (Sickle cell trait) Carriers.

Epidemiology →

- Africans with Sickle cell trait have natural protection against Falciparum malariae - not HbS.

Pathophysiology

When Sickle Haemoglobin carries oxygen its normal.
↓
When deoxygenated → Sickling (Jactoids).
↓
When recurrent causes permanent sickling
↓
Weakened cell membrane.
↓
Premature destruction → Intravascular Haemolysis
↓ Haptoglobin Jaundice Gallstone.
Bone marrow increased production
↓
"Hair on End appearance" Enlarged Bones & Skulls. Hepatomegaly

Clinical Features

- Usually precipitated by hypoxia, acidosis, dehydration and infection.

Vaso-Occlusion → Bones of hands & Feet.
↓ → Dactylitis
→ Painful Crisis

Splenic sequestration → Frequent fibrosis by infract
↓ ↳ Auto splenectomy

- Susceptible to encapsulated bacteria.
- Cerebral → stroke,
  ↳ "Moya Moya" disease → Collateral Vessels form looking like Puff of smoke"
- Blood vessel of lungs → Acute chest Syndrome.
  ↳ Recurrent Hypoxia → Recurrent sickling → Death.
- Renal Papilla → Haematuria & Proteinuria.
- Penis → Painful priapism.

Investigations:
- CBC → Anemia ↓ Haptoglobulin, ↑ Bilirubin.
→ X-ray Skull - Hair on End appearance
- Protein Electrophoresis - HbS.
- New Born are always screened
  ↳ Aminocentesis, chorionic villi

Management:
- Avoid Consanguinous marriage.

Howel Jolley Bodies
- Basophilic Nuclear Remnants in the RBC that are seen in Blood Smear.