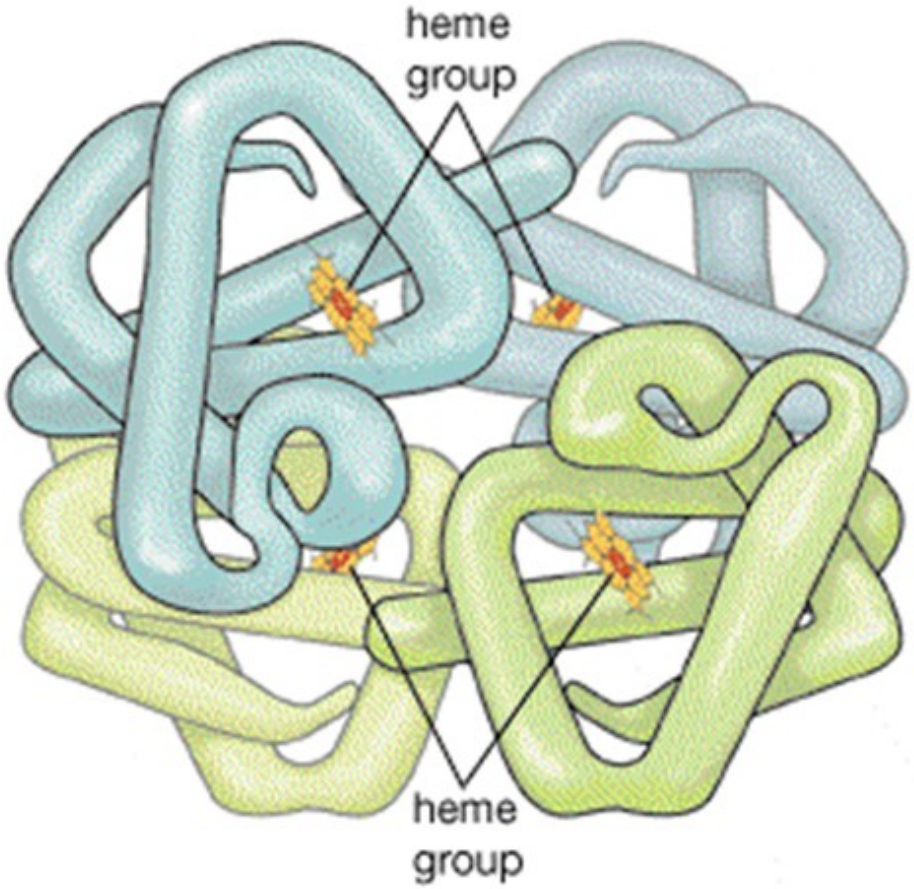
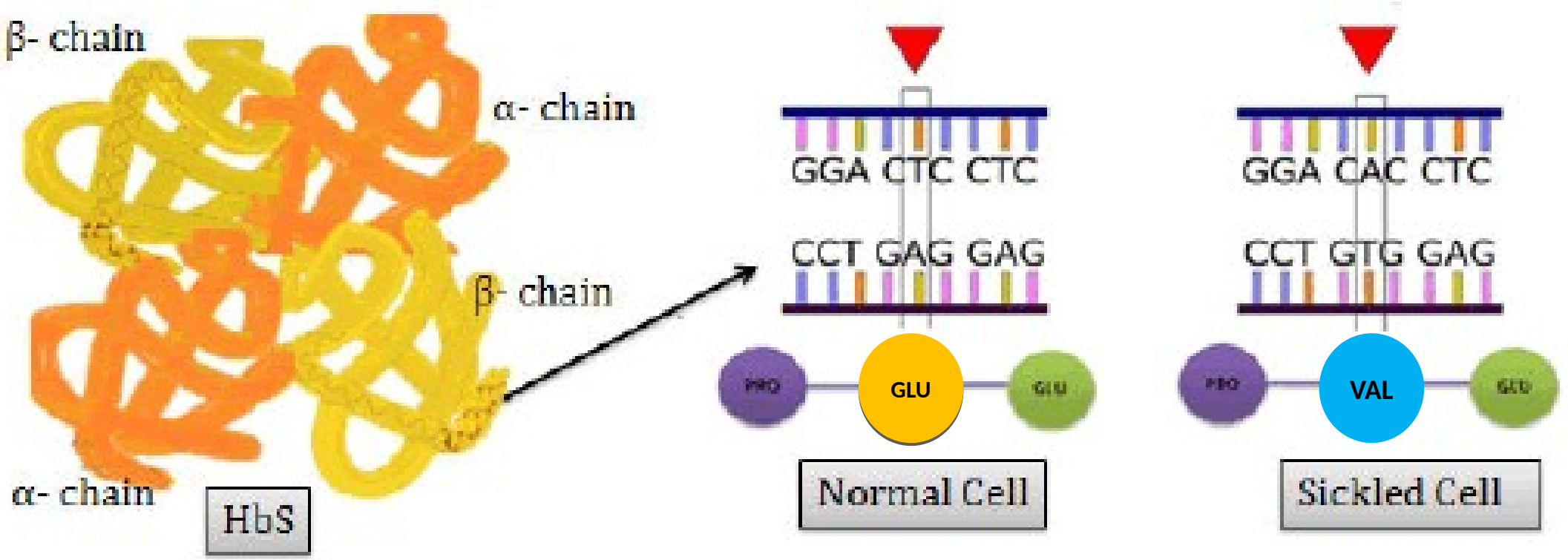
SICKLE CELL ANEMIA

DR. FAWAD RAHIM



Hb	GLOBIN SUBUNITS	DESIGNATED AS	%
A	2 Alpha – 2 Beta	α2β2	95 – 98
A2	2 Alpha – 2 Delta	α2δ2	2 – 3
F	2 Alpha – 2 Gamma	α2γ2	< 2

THE SICKLE MUTATION



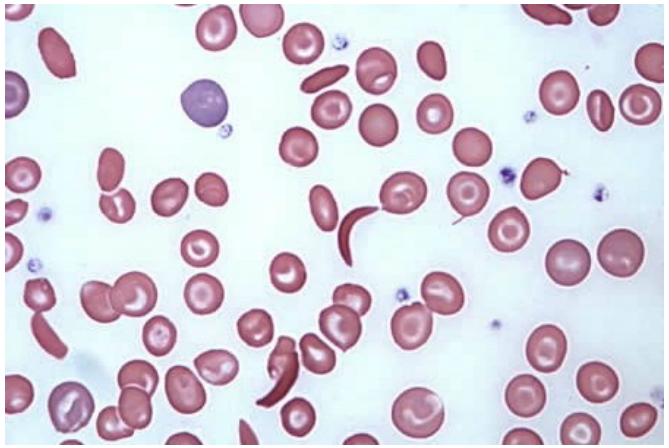
AUTOSOMAL RECESSIVE

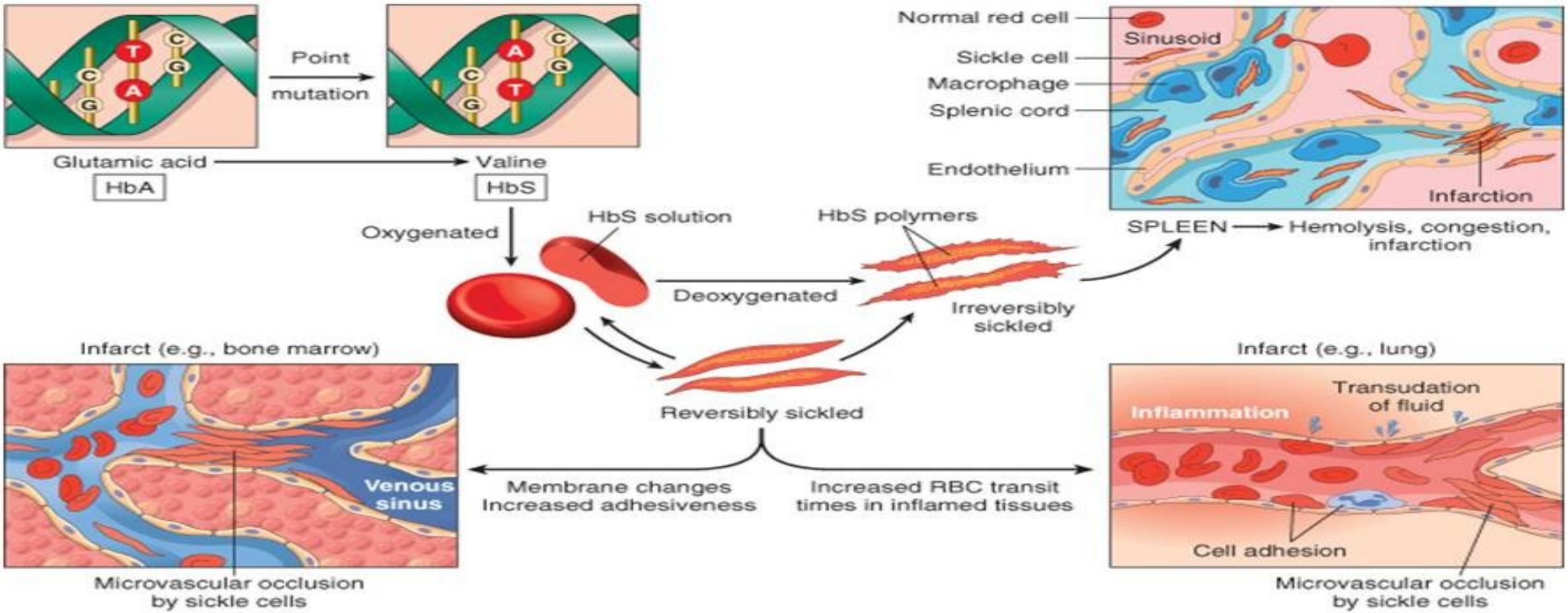
Sickle cell disease

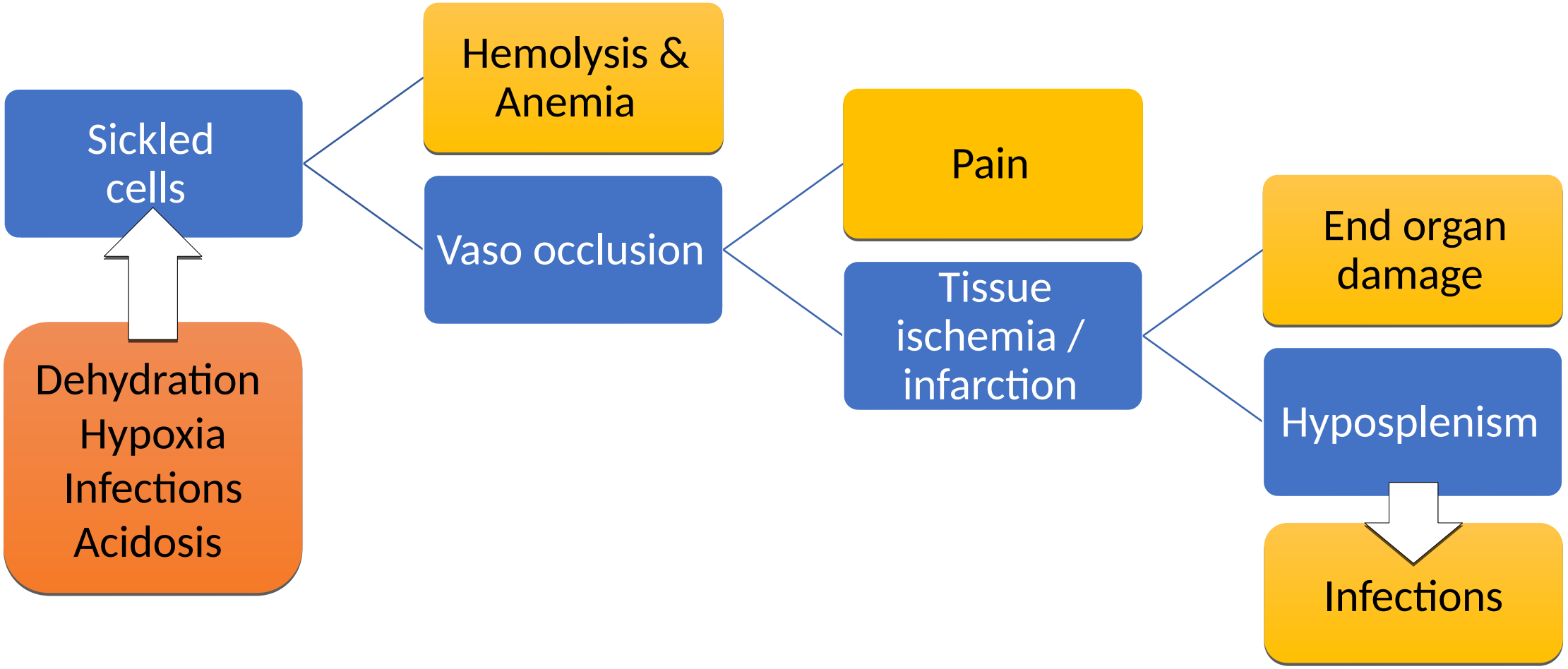
- Sickle cell anemia
- Sickle Beta Thalassemia
- Hb SC disease

Globin subunits	2 Alpha – 2 Sickle globin chains
Hb A	0 %
Hb A2	< 3.5%
Hb F	5 – 15%
Hb S	**85 – 95%**

Hemoglobin S is unstable and polymerizes in the setting of various stressors resulting in sickled RBCs






Sickled cells
Dehydration
Hypoxia
Infections
Acidosis
Hemolysis & Anemia
Vaso occlusion
Pain
Tissue ischemia / infarction
End organ damage
Hyposplenism
Infections

ANEMIA

- Chronic compensated hemolytic anemia
- Aplastic crisis
- Splenic sequestration crisis
- Hyperhemolytic crisis

PAIN

Acute painful episodes

- Acute chest syndrome
- Acute multi-organ failure
- Acute surgical abdomen
- Acute papillary necrosis
- Acute splenic or hepatic sequestration crises
- Opioid withdrawal
- Acute coronary syndrome
- Osteomyelitis

Chronic pain

END ORGAN DAMAGE

ACUTE

- Stroke
- Acute chest syndrome
- Renal infarction
- Dactylitis or bone infarction
- Myocardial infarction
- Priapism
- Venous thromboembolism

CHRONIC

- Anemia
- Neurologic deficits or seizure disorder
- Pulmonary hypertension
- Renal impairment and hypertension
- Osteoporosis and complications of bone infarction
- Cardiomyopathy with diastolic dysfunction
- Hepatotoxicity and pigmented gallstones
- Delayed puberty and reduced growth
- Chronic leg ulcers
- Proliferative retinopathy

INFECTIONS

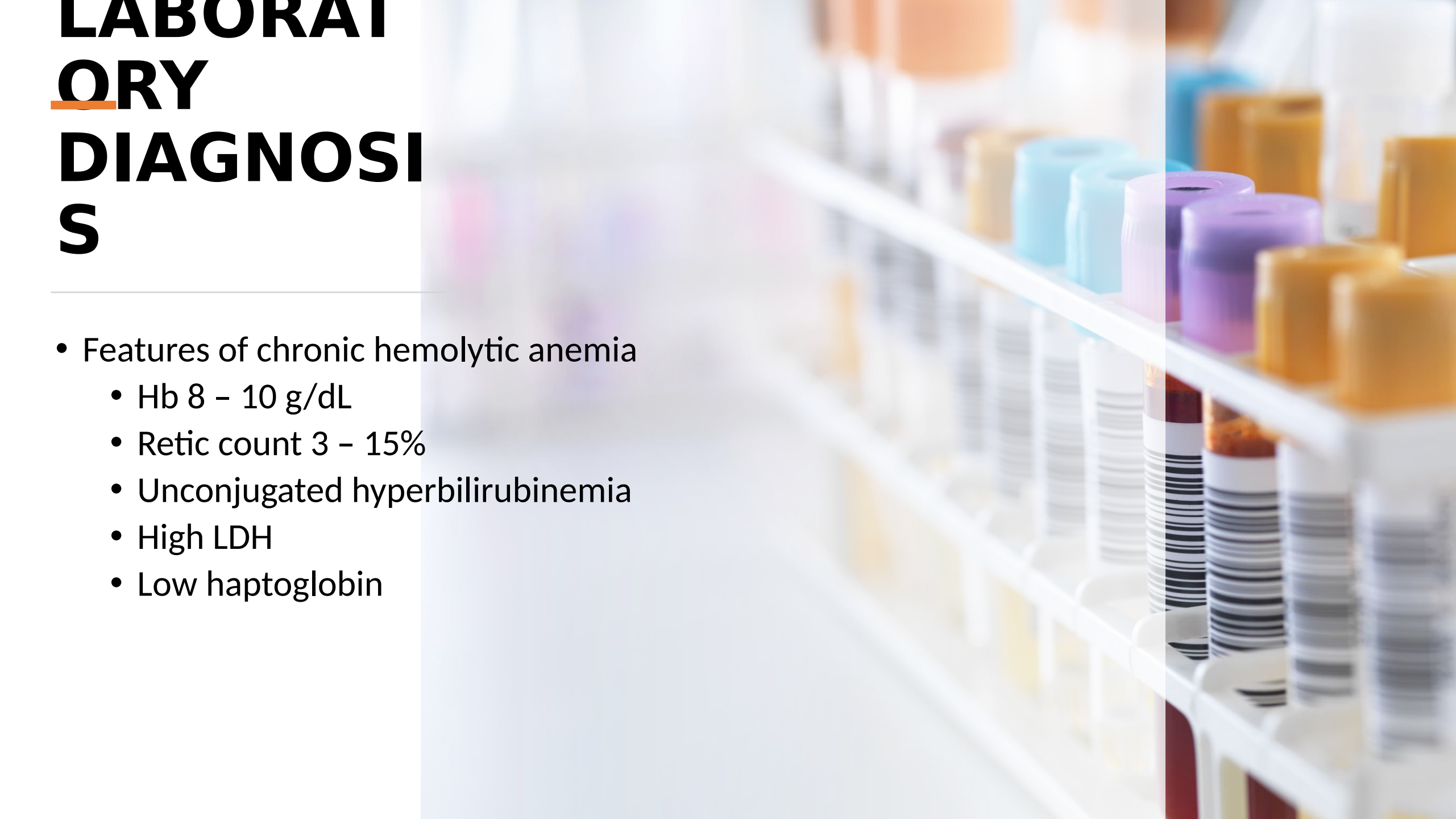
- Bacteremia
- Meningitis
- Pneumonia

PHYSICAL EXAMINATION

- Chronically ill, stunted growth
- Pallor
- Jaundice
- Hepatomegaly
- Enlarged heart ±
- Non healing leg ulcers
- Retinopathy

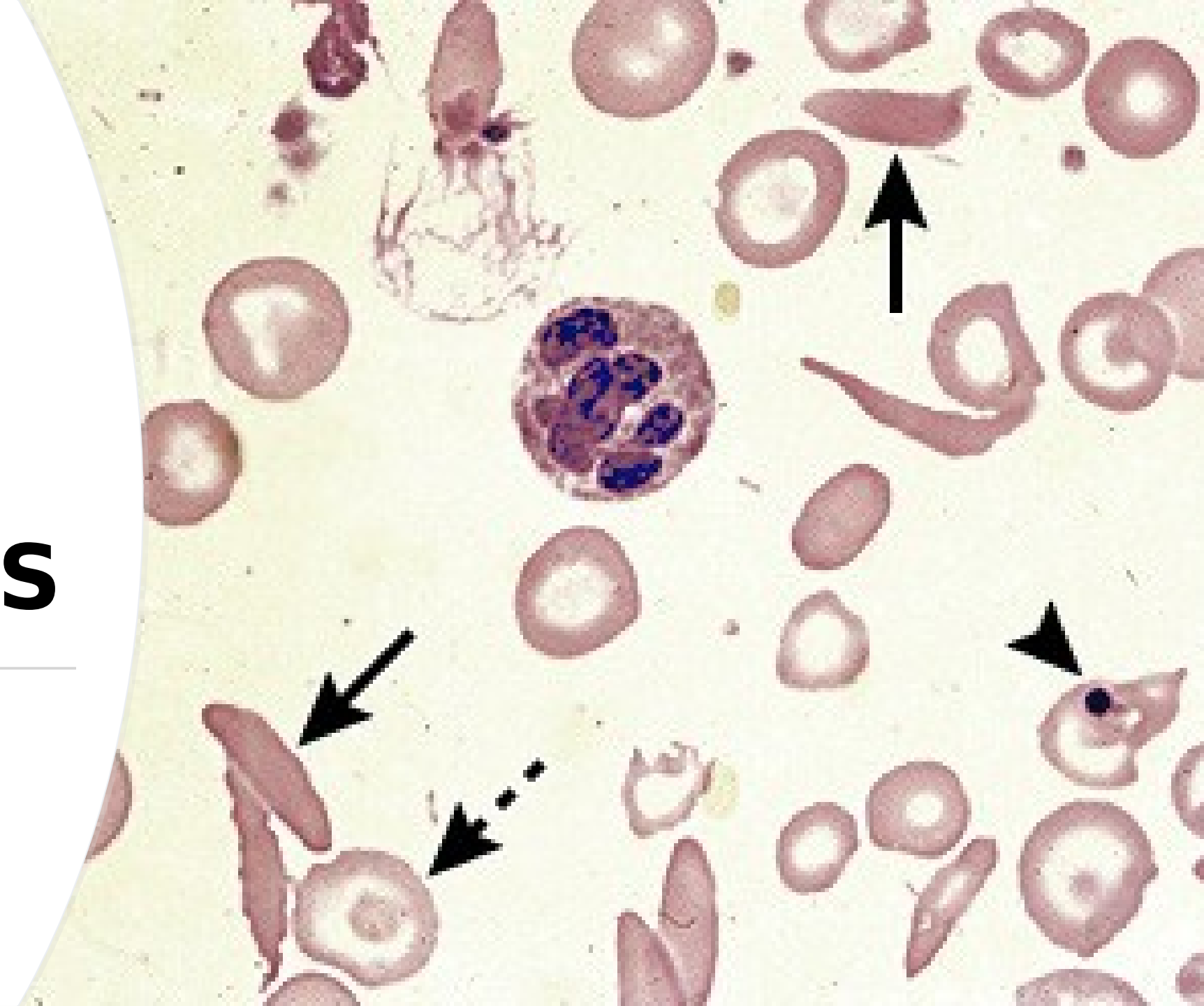
LABORATORY DIAGNOSIS

- Features of chronic hemolytic anemia
  - Hb 8 – 10 g/dL
  - Retic count 3 – 15%
  - Unconjugated hyperbilirubinemia
  - High LDH
  - Low haptoglobin



LAB DIAGNOSIS

Peripheral smear



HB ELECTROPHORESIS

- Diagnostic test for sickle cell anemia

Globin subunits	2 Alpha – 2 Sickle globin chains
Hb A	0 %
Hb A2	< 3.5%
Hb F	5 – 15%
Hb S	**85 – 95%**

MANAGEMENT

Supportive care

- Folic acid
- Transfusion
- Acute painful episodes
- Vaccination

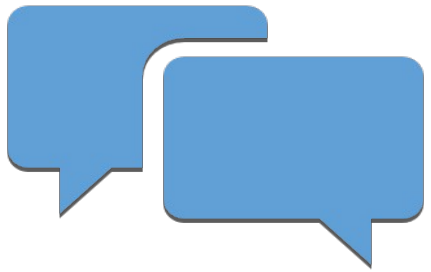
Cytotoxic agents e.g. Hydroxyurea

Inhibitors of sickling - voxelotor

Inhibitors of endothelial adhesion - crizanlizumab

Exchange transfusion

Allogenic hematopoietic stem cell transplant



Comments

Queries