HLS - LAB

Dr.Eman Kreishan, M.D.

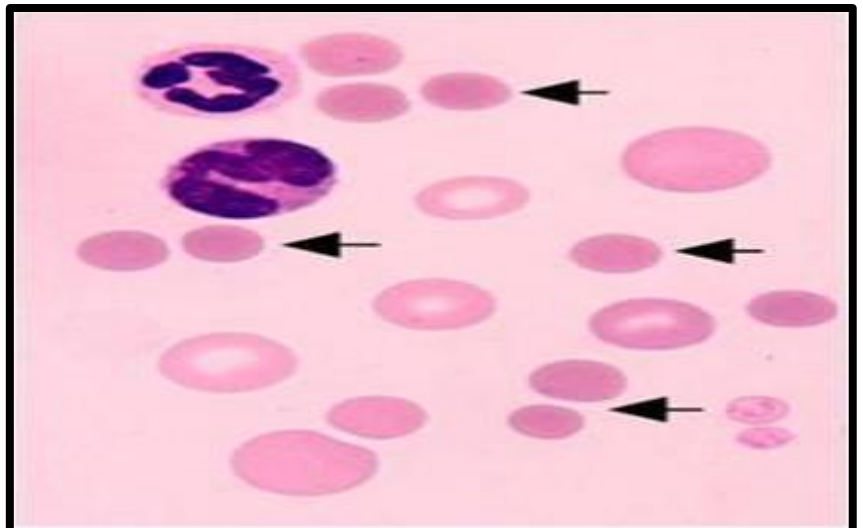
HEREDITARY SPHEROCYTOSIS

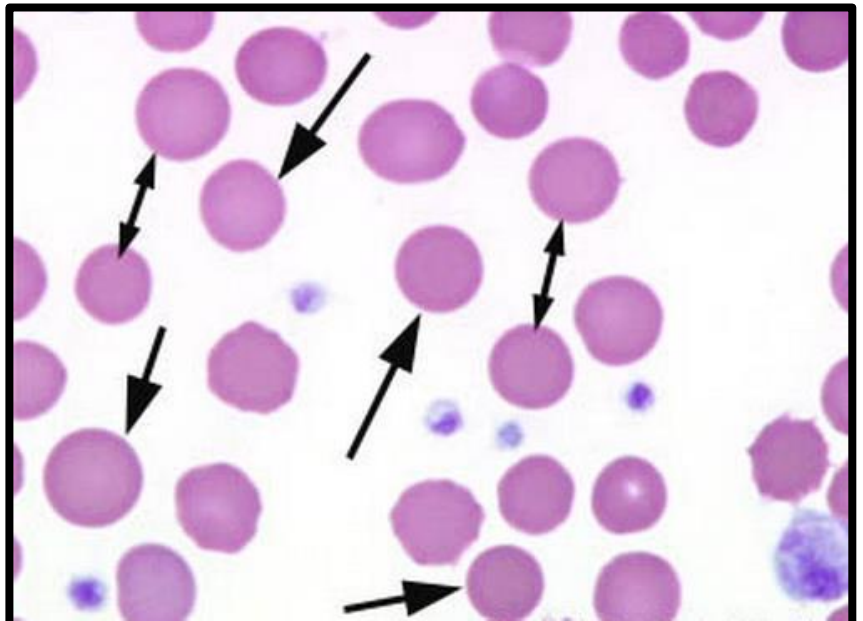
Usually transmitted as Autosomal dominant trait, due to mutations in Spectrin, Ankyrin, and band 3.

Clinically characterized by:

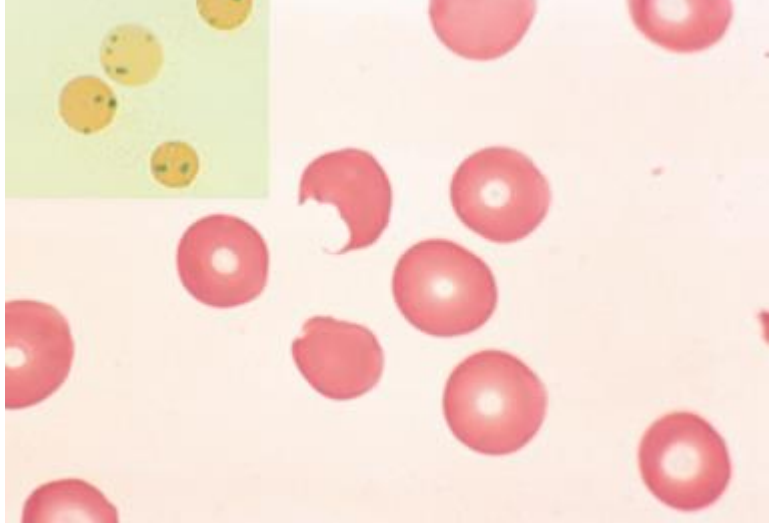
(Severity of anemia correlates with spectrin deficiency):

- Anemia, splenomegaly and jaundice.
- Gallbladder stones
- Aplastic crisis: Due to infection by Parvovirus B19





G6PD Deficiency



- Because regeneration of GSH is impaired in G6PD-deficient cells.
- oxidants are free to “attack” red cell components including globin chains.
- Oxidized hemoglobin denatures and precipitates, forming Heinz bodies, which can damage the red cell membrane (intravascular hemolysis).
- Other cells with lesser damage lose their deformability and suffer further injury when splenic phagocytes attempt to remove the Heinz bodies, creating bite cells, (extravascular hemolysis).

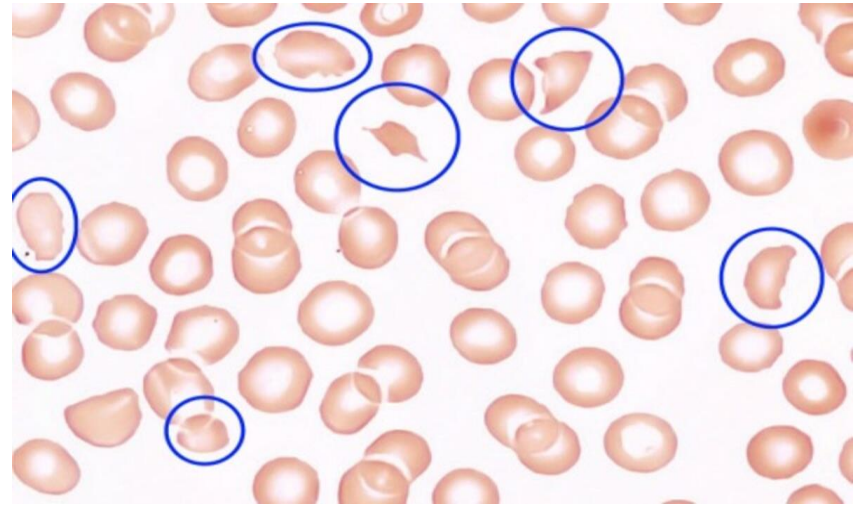
Hemolytic Anemia Due to Mechanical Trauma to RBC

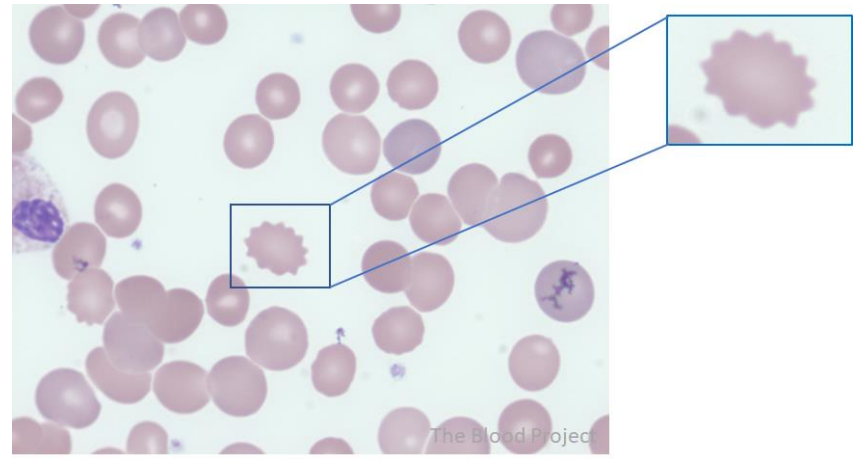
Etiology

- Artificial valves
- Microangiopathic hemolytic anemia
  - DIC
  - Malignant hypertension
  - TTP
  - Hemolytic uremic syndrome

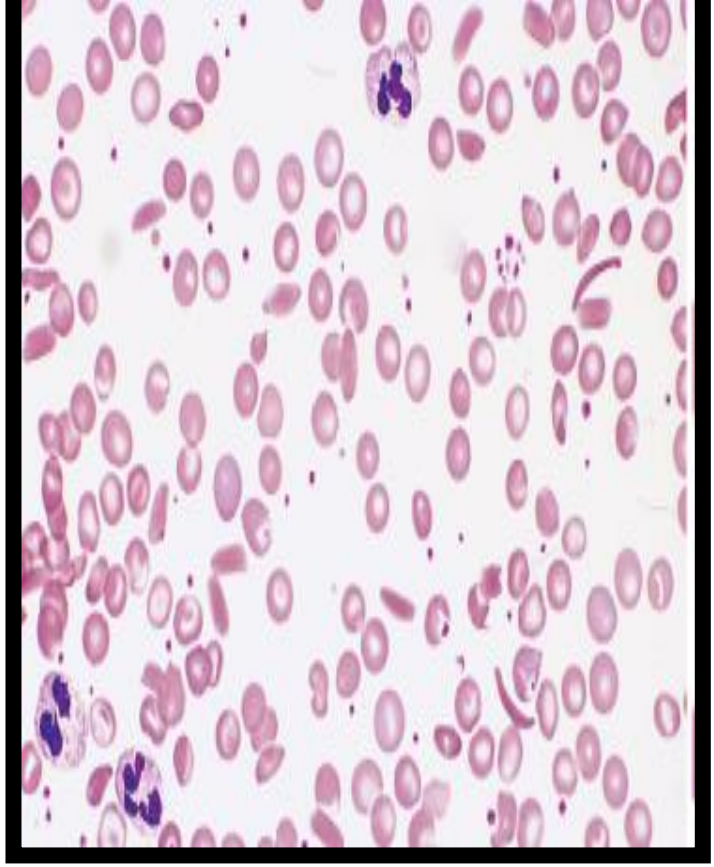
Morphology

- Significant poikilocytosis with helmet cells/ schistocytes , burr cells, and triangle cells





SICKLE CELL DISEASE

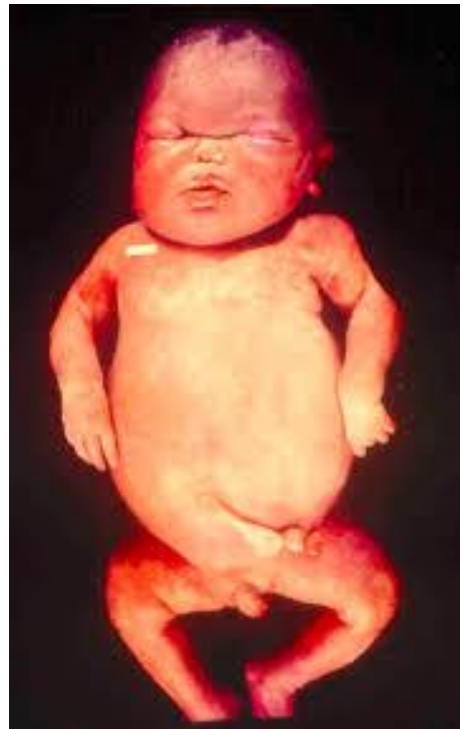


- Asymptomatic till 6 months of age.
- Moderate to severe anemia (6-8 g/dl).
- Unremitting course complicated by sudden crises.
- Laboratory investigation: CBC and blood smear, Hemoglobin electrophoresis.
- Treatment: Adequate hydration / Pain relief/ Antibiotic therapy/ exchange transfusion to reduce the HbS.

α-Thalassemia

- Caused mainly by deletions involving one or more of the a-globin genes.
- Severity of the disease is proportional to the number of a-globin genes that are missing.

- -α/αα: silent carrier state: asymptomatic
- --/αα, -α/-α : α thalassemia minor: asymptomatic
- --/-α : Excess beta: Beta 4: HbH disease
- --/-- : Excess Gamma : Hb Barts, Death in utero
- (Hydrops fetalis)



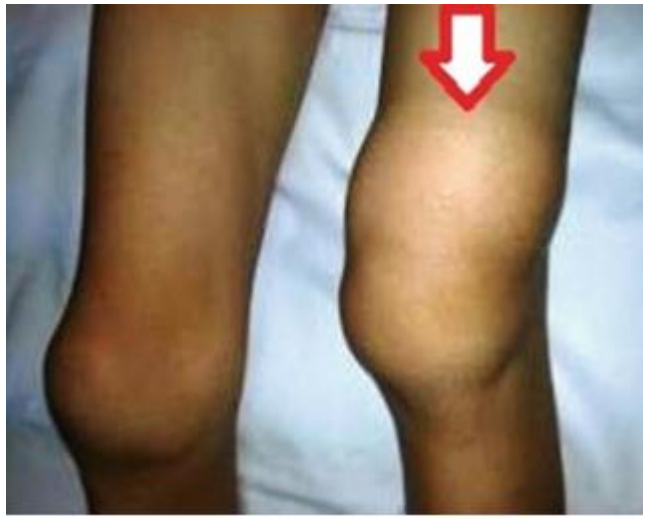
I. Hemophilia A

- Hemophilia A is an X-linked, recessive disorder caused by deficiency of functional plasma clotting factor VIII (FVIII).

- Hemophilia A is the most common X-linked genetic disease and the second most common factor deficiency after von Willebrand disease (vWD).

- Occurs predominantly in males, Females usual asymptomatic carriers, but??????????????.

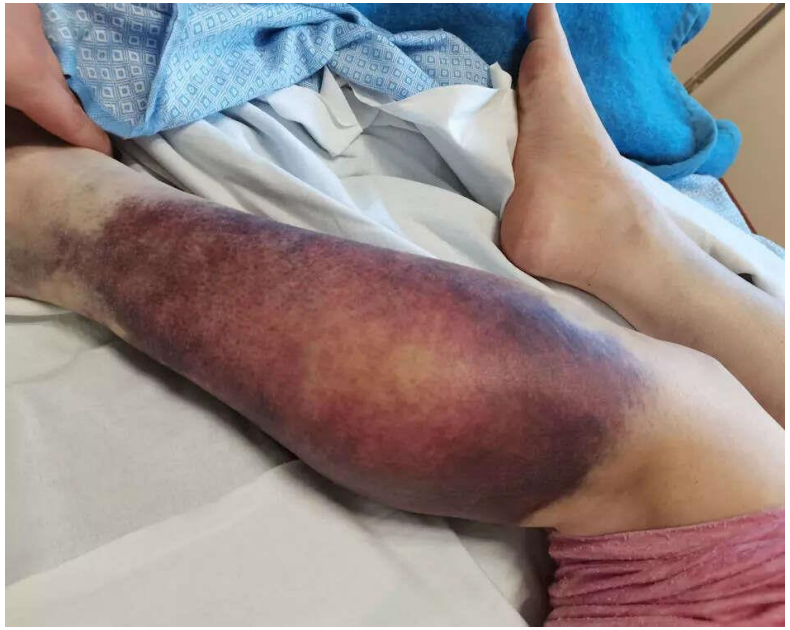
Signs and symptoms



- melena.
- Hemarthrosis
- Hematemesis
- oral mucosal hemorrhage
- Epistaxis
- Hematuria.

- Clinical presentation: Same as Hemophilia A.

- Laboratory tests:
  - Prolonged PTT. Normal PT and TT.
  - normal factor VIII assay.



- Treatment of hemophilia may involve:
  - Management of bleeding episodes.
  - Use of factor replacement products and medications.
  - Rehabilitation of patients with hemophilic synovitis.

von Willebrand disease

✓The most common hereditary bleeding disorder .

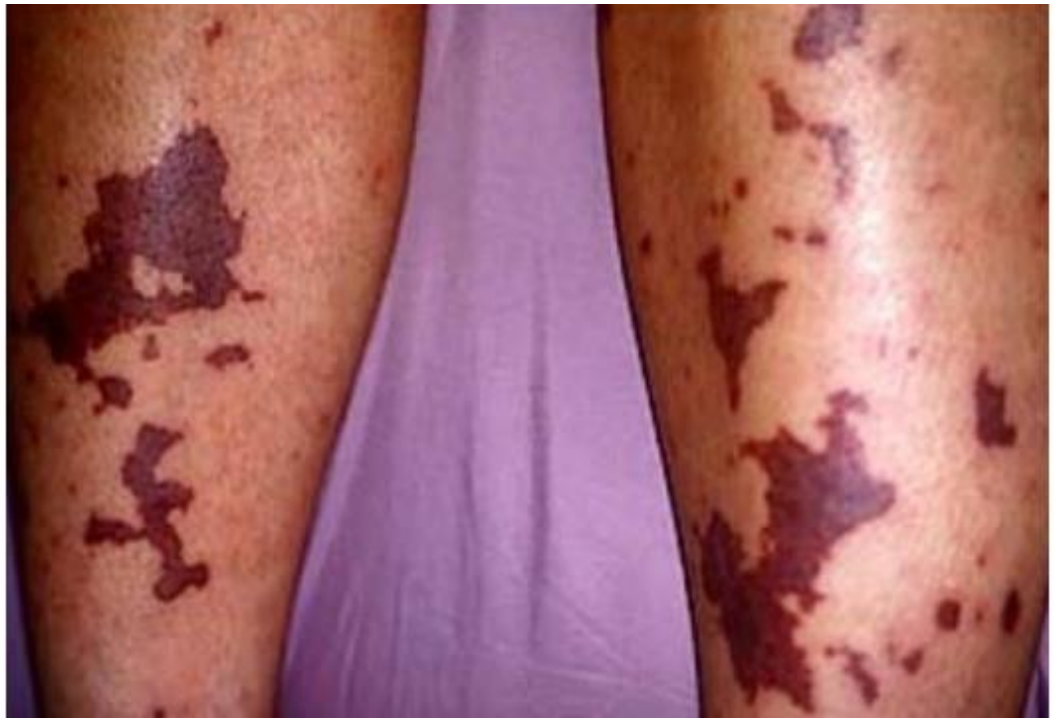
✓von Willebrand disease is transmitted as an autosomal dominant disorder.

✓Presented with mild bleeding problems such as:

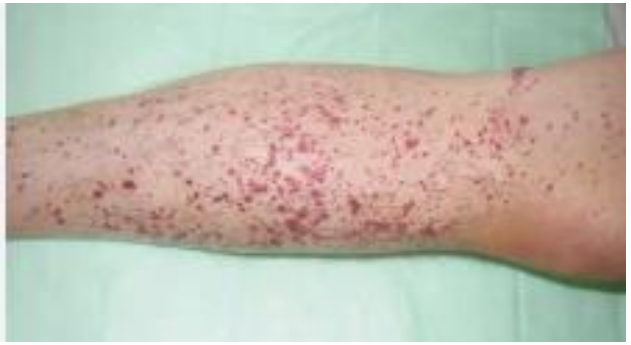
- Mucous membrane bleeding
- Easy bruising
- menorrhagia
- Post-operative bleeding.

*Both sexes are affected, and presented with prolonged bleeding times (BT) despite normal platelet counts.

DIC (Consumptive Coagulopathy)



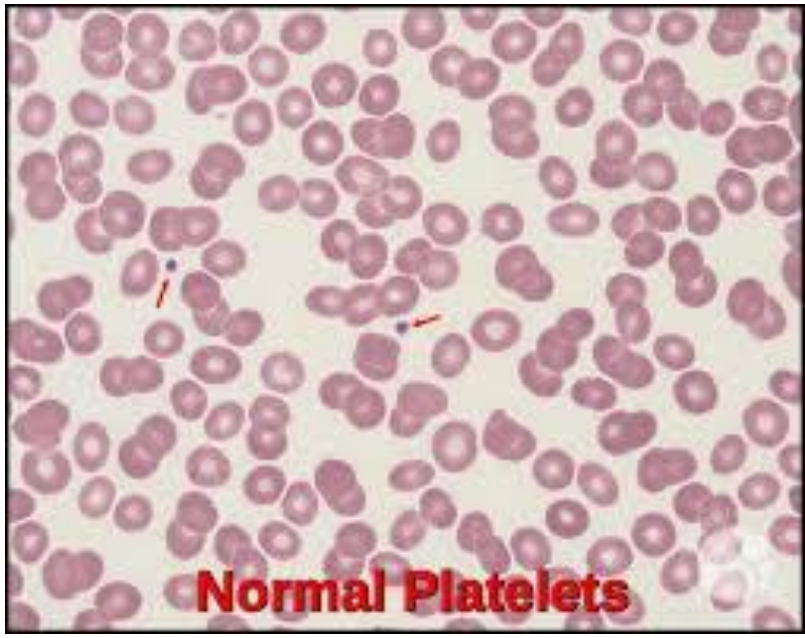
Thrombocytopenia

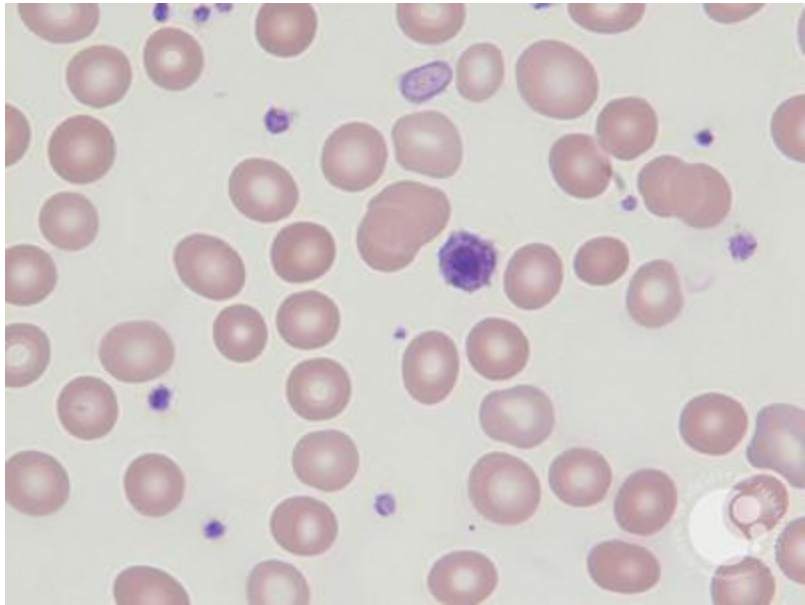


- Normal range 150-450X10^3 per µl.
- Levels above 60x10^3/µl will not cause bleeding under normal conditions.
- Levels below <u>20x10^3/µl</u> will cause:
  - Petechiae, mucosal bleeding.
  - Post-operative bleeding, CNS bleeding.
- Levels around 5x10^3/µl can lead to fatal CNS or GI hemorrhage.
- Levels between <u>20 and 60x10^3/µl</u> may cause bleeding (depending on platelets functional status).

Immune thrombocytopenia (ITP)

- * On CBC:
- Decreased platelet count (10-50x109/l), normal Hb and WBCs.
- * Peripheral blood: large platelet.
- * Bone marrow: Increased Megakaryocytes numbers.
- * Bleeding time: : Mild prolongation.
- * Assay for Antiplatelet antibodies.





Thrombotic thrombocytopenic purpura

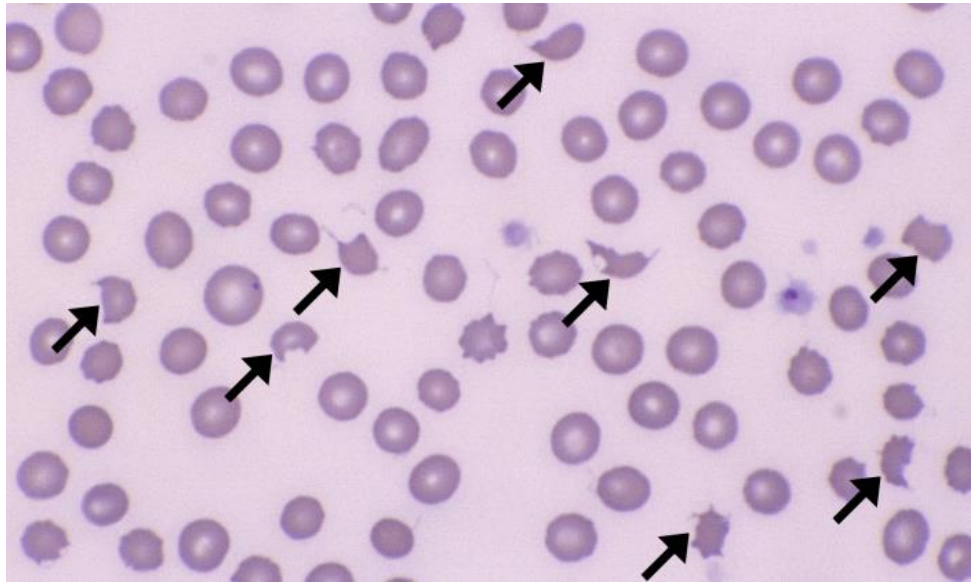
- *#Laboratory studies for suspected TTP include:*

➢ *CBC with platelet count.*

➢ *peripheral blood smear.*

➢ *coagulation studies(*Normal PT, PTT, D-Dimer but elevated BT).

➢ **Signs of hemolysis:** Increase LDH, Increase indirect bilirubin Decrease Haptoglobin

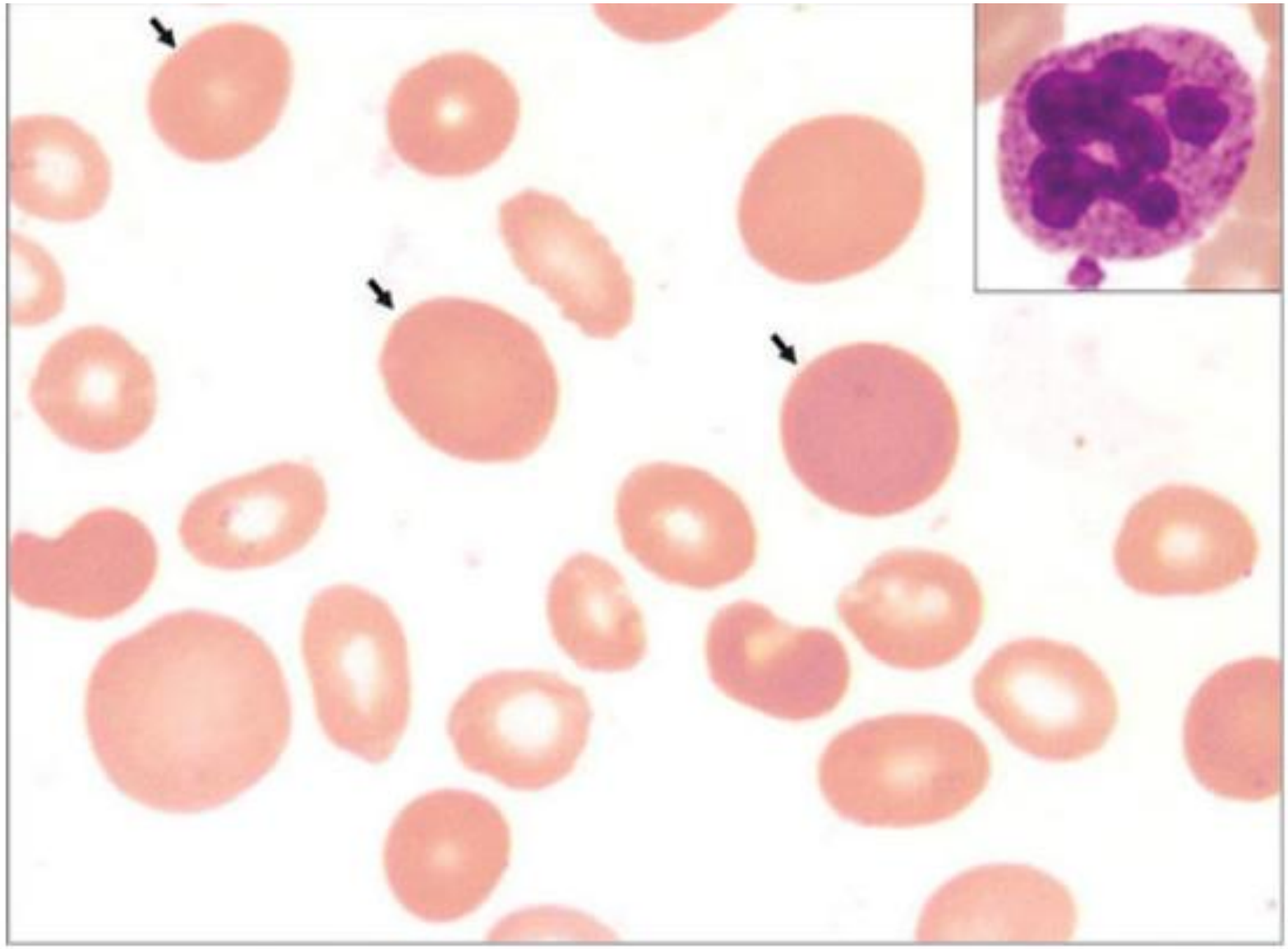


➢ *BUN and creatinine.*

➢ Measuring ADAMTS13 activity level.

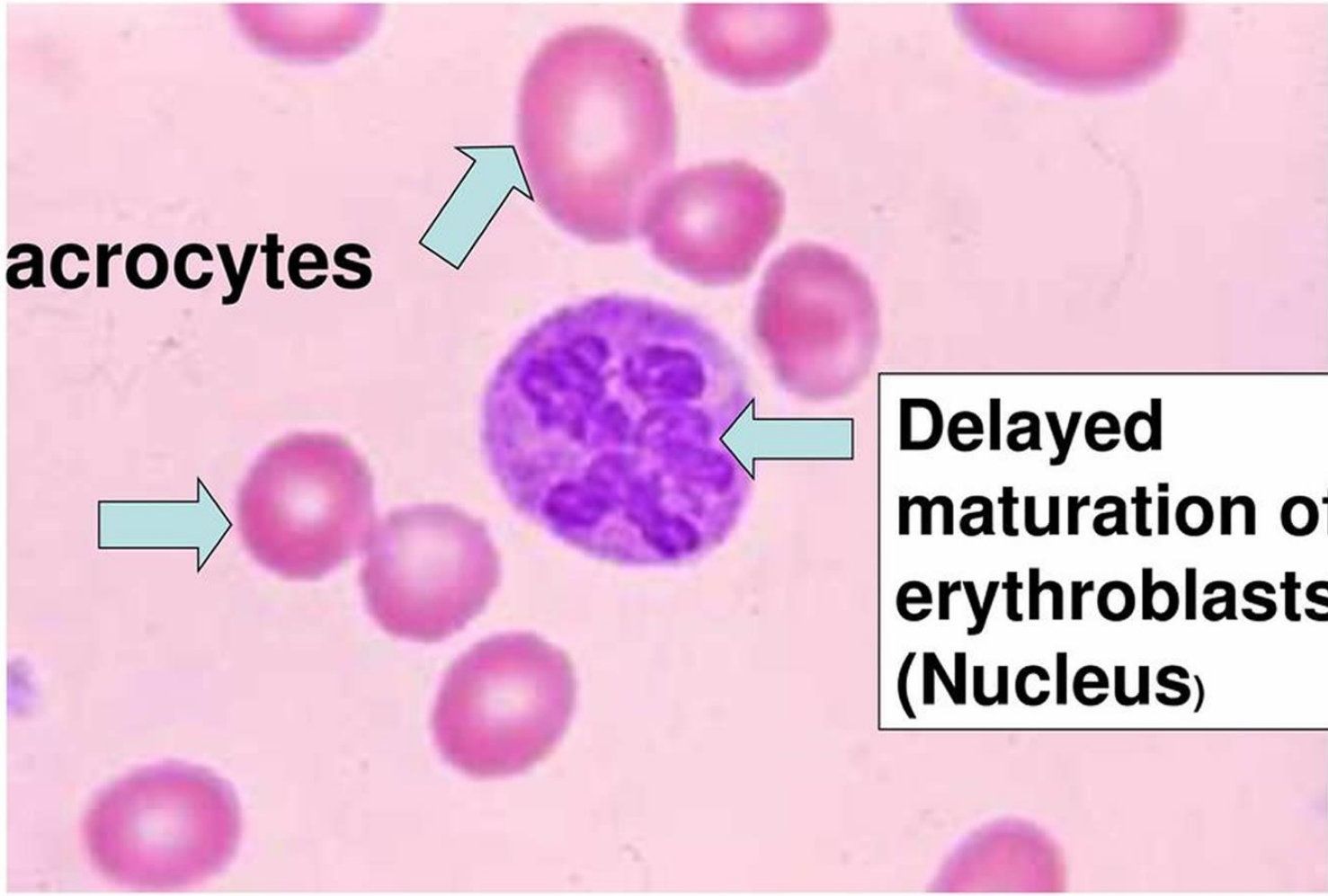
➢ Treatment:

- The therapy of choice for TTP is plasma exchange with fresh frozen plasma



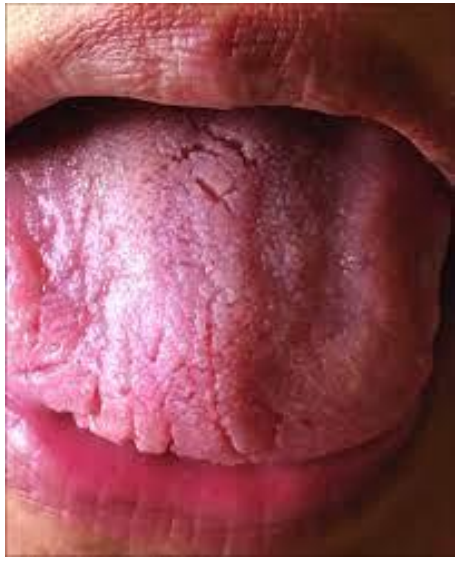
. 1.3: Peripheral blood smear showing macro-ovalocytes (arrows) and hypersegmented neutrophil (inset)

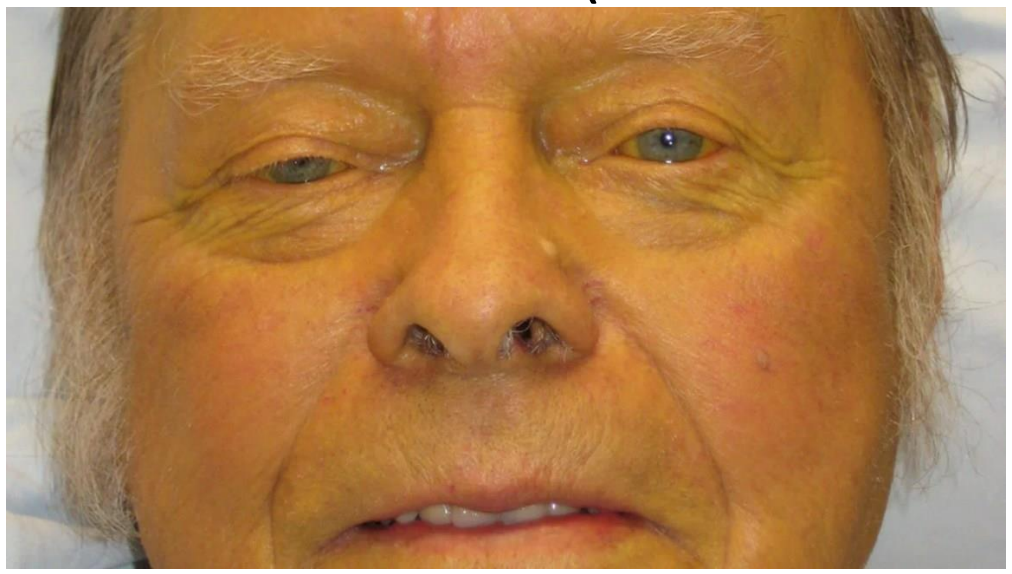
Megaloblastic Anemia

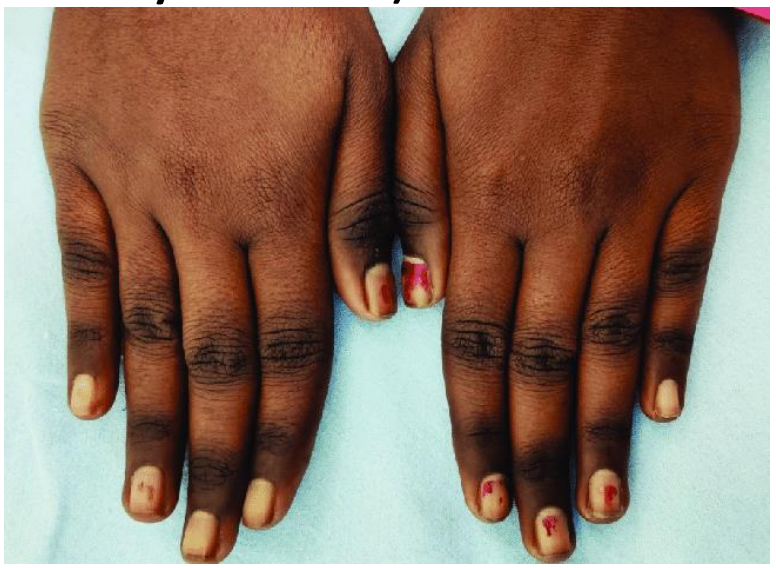


Megaloblastic anemia

- * Glossitis, characterized by a smooth tongue due to loss of papillae, occurs in persons with cobalamin deficiency.
- * Patients may have a lemon-yellow hue due increased indirect bilirubin level (intramedullary hemolysis).
- * hyperpigmentation of the skin (increased melanin synthesis).







Iron def anemia

- Fatigue and diminished capability to perform hard labor.
- Leg cramps on climbing stairs.
- Cold intolerance.
- abnormalities of the fingernails (thinning, flattening, and “spooning,”) and pica

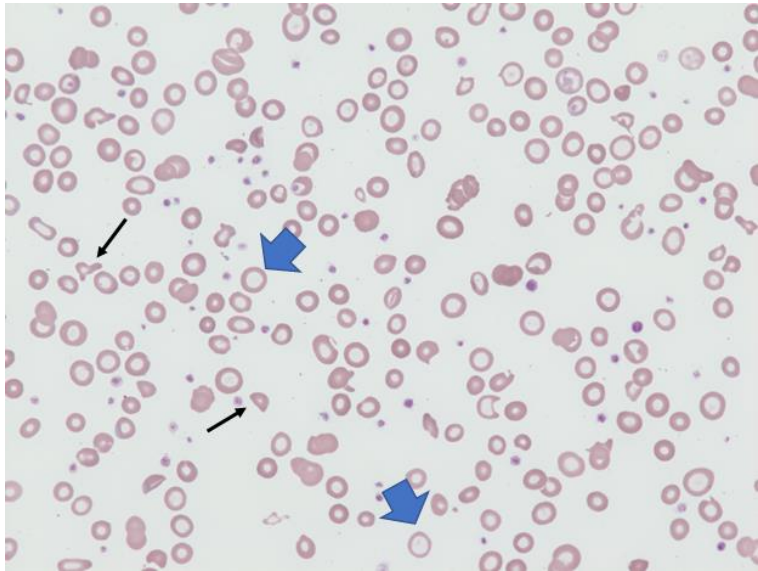
Laboratory manifestation

❑Complete blood count (CBC):

- microcytic and hypochromic erythropoiesis, decrease in both mean corpuscular volume (MCV) and the mean corpuscular hemoglobin concentration (MCHC).

❑Low serum iron and ferritin levels

❑Peripheral Smear: microcytic and hypochromic red



Aplastic anemia

- Aplastic anemia is a syndrome of bone marrow failure characterized by peripheral pancytopenia and marrow hypoplasia