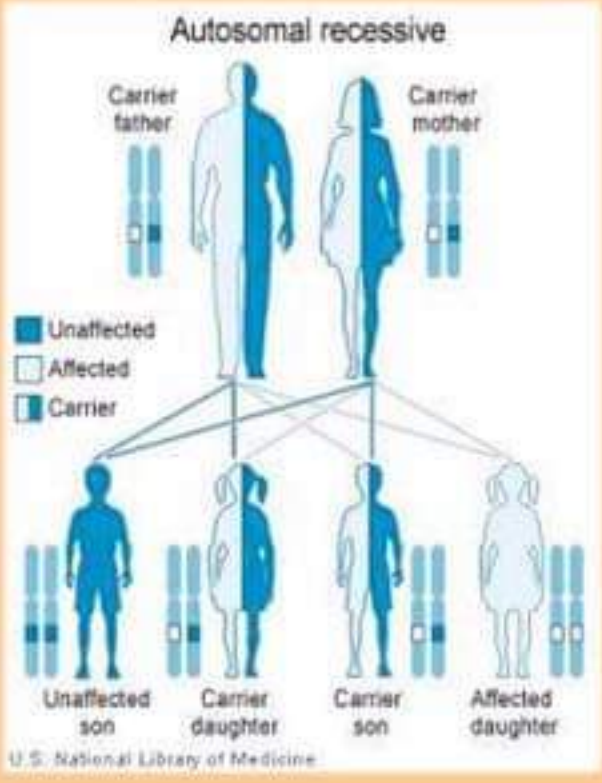
Thalassemia

A blood disorder passed down through families in which the body makes an abnormal form of hemoglobin.
This results in excessive destruction of red blood cells, which leads to anemia.
Occurs when there is a defect in a gene that helps control production of one of these proteins (Alpha globin and Beta globin)

Symptoms

- Bone deformities in the face
- Fatigue
- Growth Failure
- Shortness of Breath
- Yellow Skin
- Swollen Spleen



Expectations

- Early death due to heart failure, usually between ages 20 and 30.
- Frequent blood transfusions with therapy to remove iron from the body help improve the outcome.

Treatment

- Blood Transfusions
- Folate Supplements
- Chelation Therapy (to remove excess iron)
- Bone Marrow Transplants

Complications

- If left untreated it can lead to:
  - Heart Failure
  - Liver Problems
  - It also makes a person more likely to develop infections

Fun Facts

- Alpha Thalassemia occurs mainly in people from southeast Asia, the Middle East, China, and those of African descent
- Beta occurs commonly in people of Mediterranean origin, and to a lesser extent, Chinese, other Asians, and African Americans

http://www.ncbi.nlm.nih.gov/pubmedhealth/PMH0001613/
https://atlasmichealth.dnadirect.com/gro/patient-site/beta-thalassemia-carrier-screening/genetics-of-beta-thalassemia.html

HISTORY

Thalassemia First described by Dr Thomas Cooley in 1925.

He observed the disorder in patients of mediteranean ancestry, and called “Cooley's anemia”. •

Doctors at the University of Rochester "Whipple and Bradford” proposed the name Thalassemia.

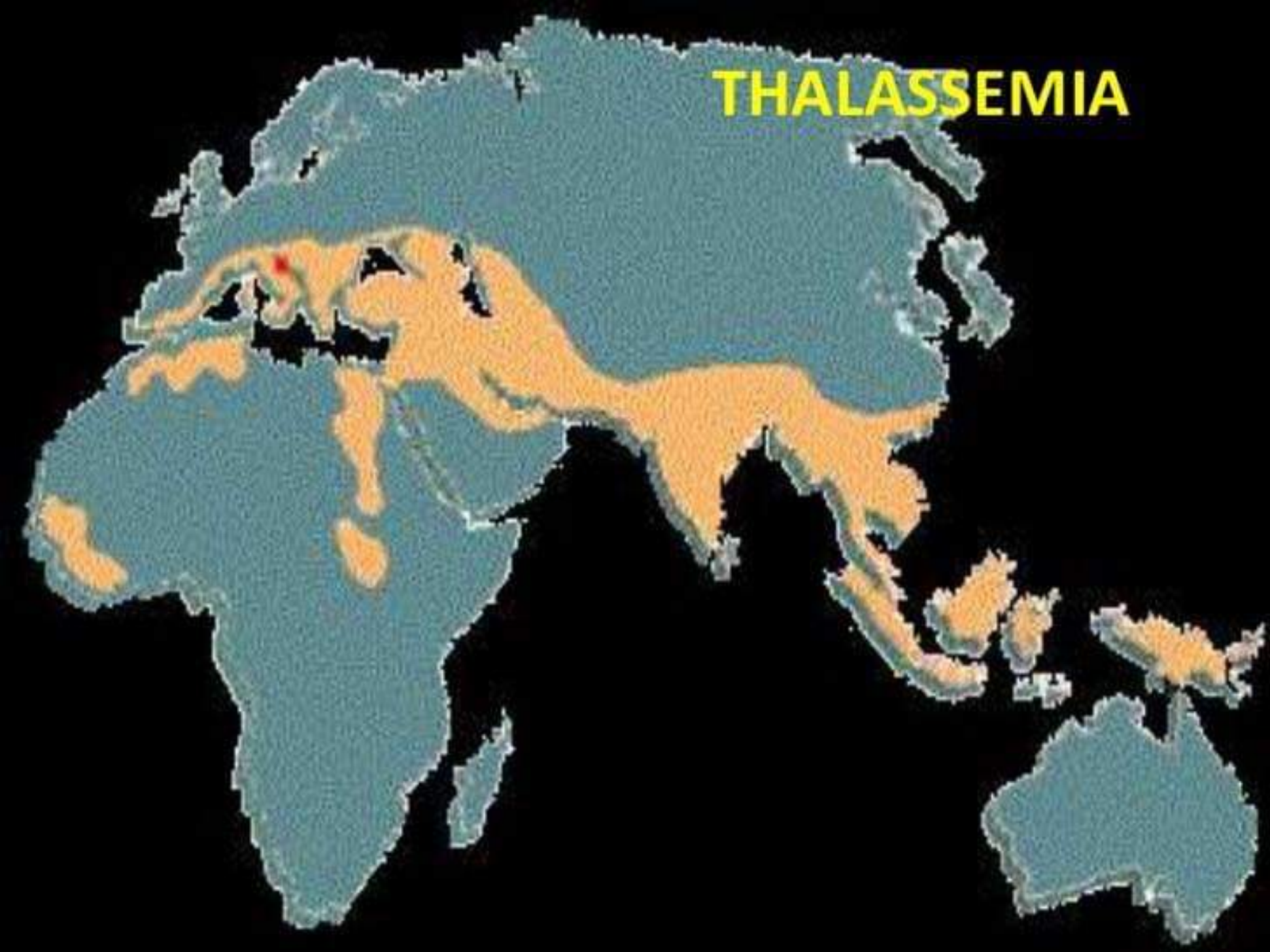
OVERVIEW

- The name is derived from the Greek word "Thalasso = Sea" and "Hemia = Blood" in reference to anemia of the sea. History of Thalassemia
- THALASSEMIA is a heterogenous group of disorders characterized by **genetically determined reduction in** the rate of **synthesis** of **normal** globin chain.
- Commonest form of haemoglobinopathy.

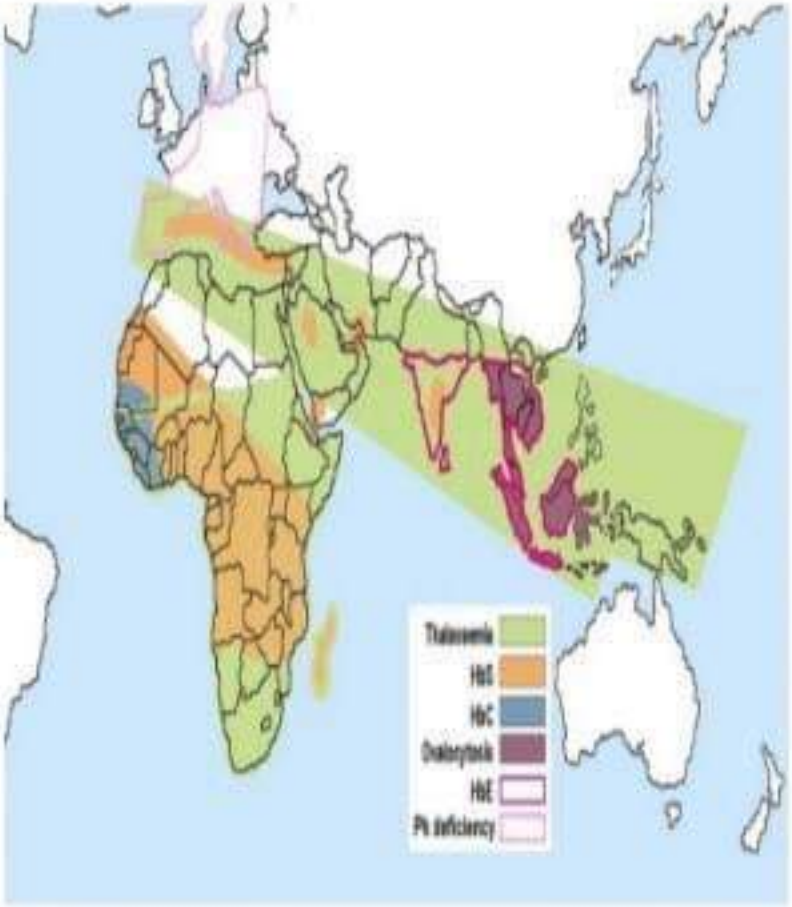
PREVALENCE

- The alpha thalassemia is prevalent in southeast Asia, Malaysia and southern china.

- The beta thalassemia are seen primarily in the area surrounding Mediterranean sea, Africa and southeast Asia.
- Carrier frequency of thalassemia in India is about 3 % and estimated frequency of thalassemia at birth is 1:2700.


THALASSEMIA

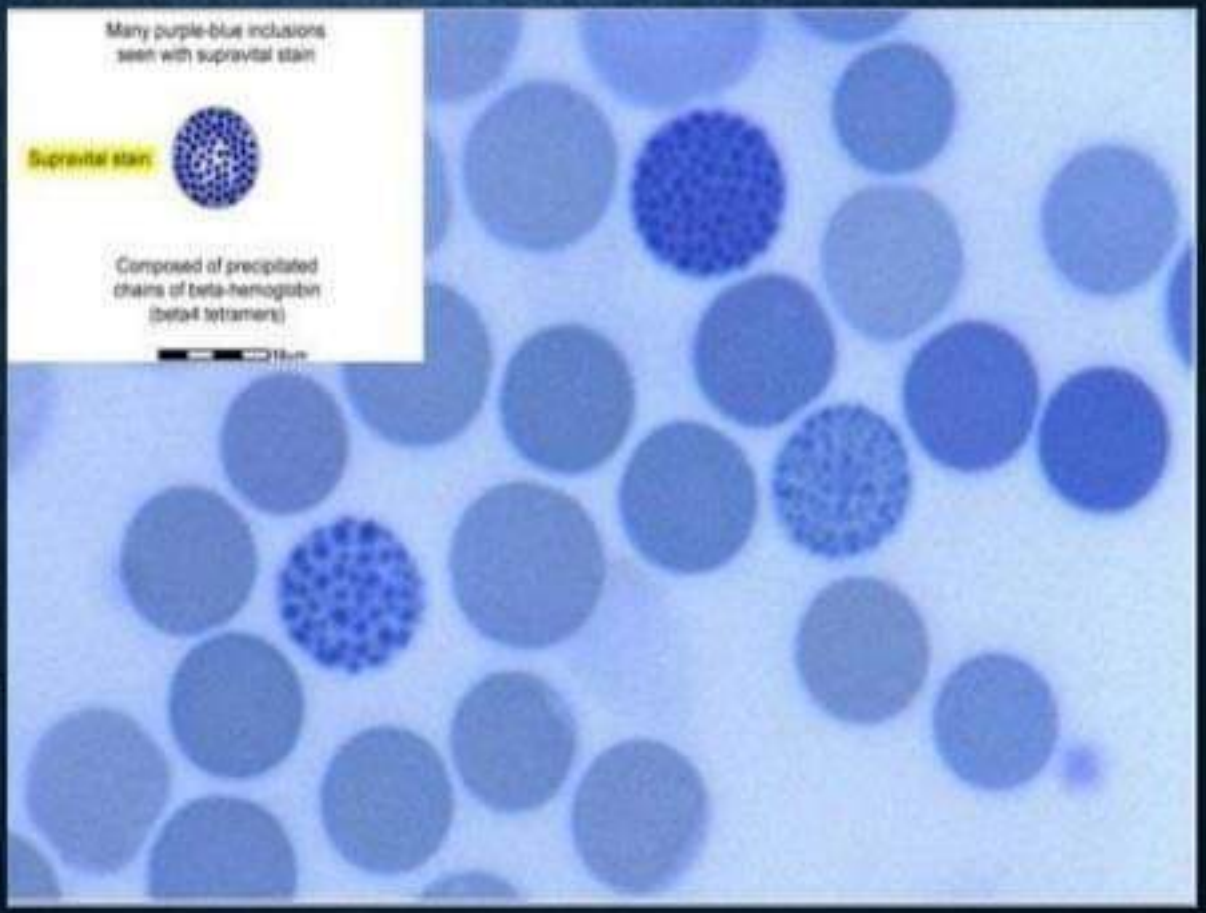
Geographical Distribution



- **Thalassemia** is found in parts of the world where malaria is common.

- It occurs most frequently in people from :
  - Mediterranean countries
  - North Africa
  - The Middle East
  - India
  - Central & Southeast Asia.

HbH Inclusion Bodies



Types

Thalassemia Major (Cooley's anemia)

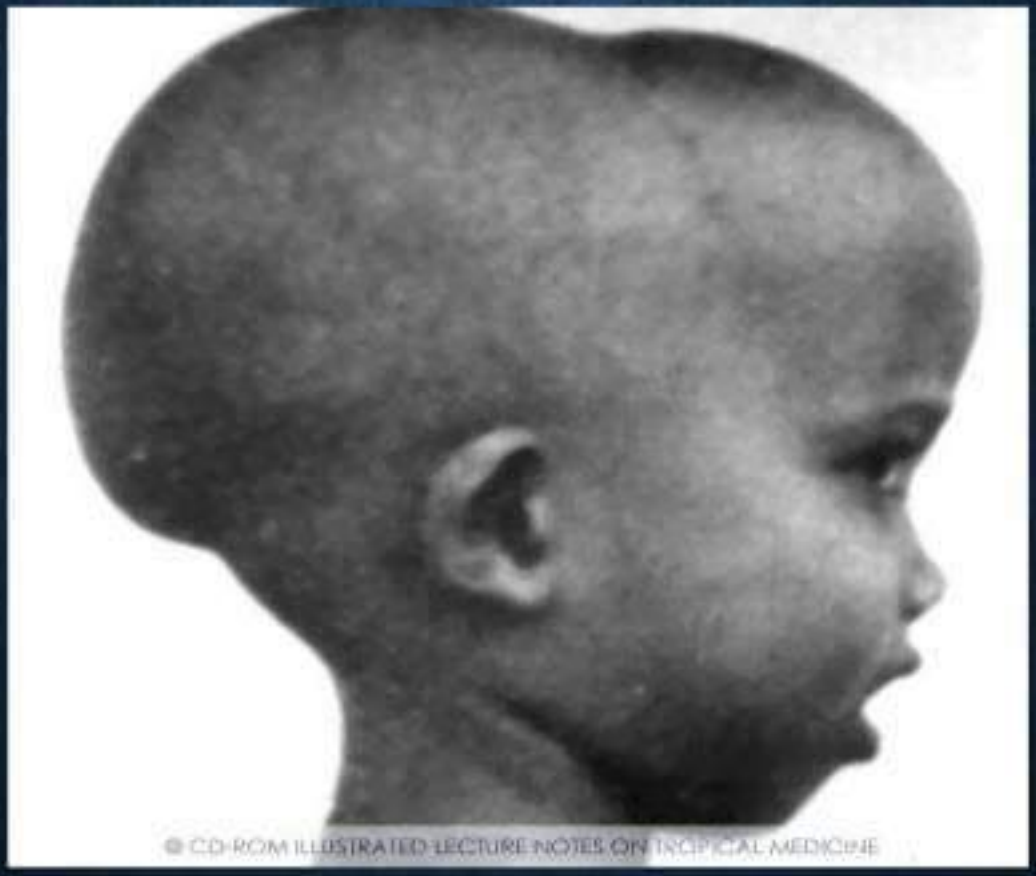
-severe form of beta thalassemia

- presence of two abnormal genes that cause either a severe decrease or complete lack of beta globin production.

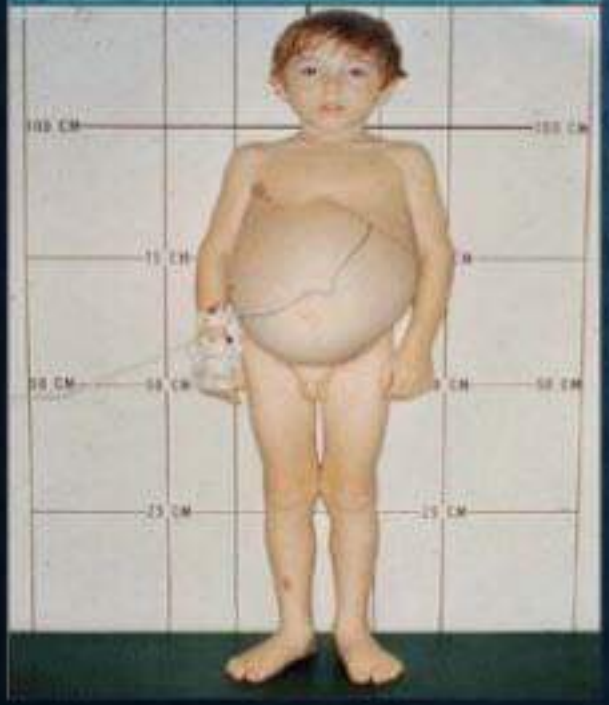
Thalassemia Minor

- presence of one normal gene and one with a mutation

- causes mild to moderate mild anemia.

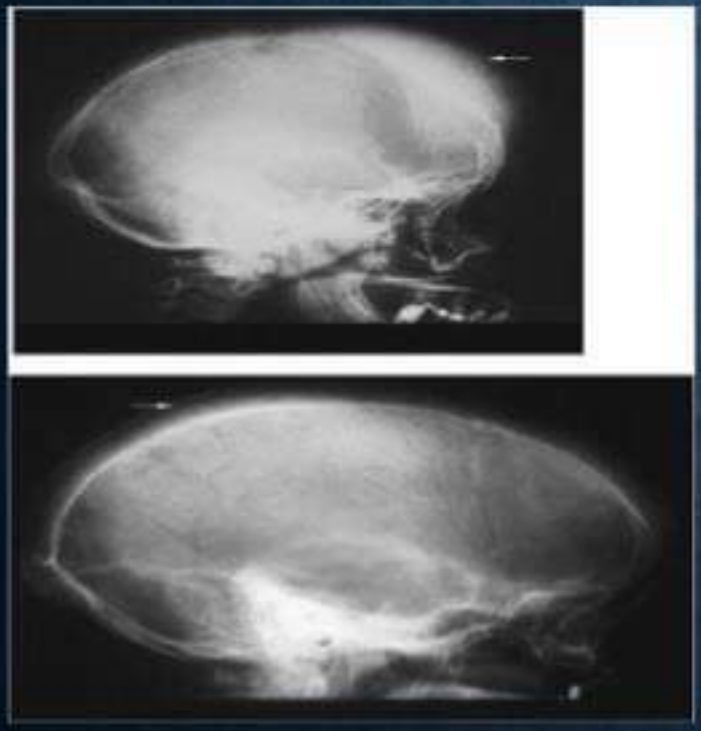

© CD-ROM ILLUSTRATED LECTURE NOTES ON TROPICAL MEDICINE

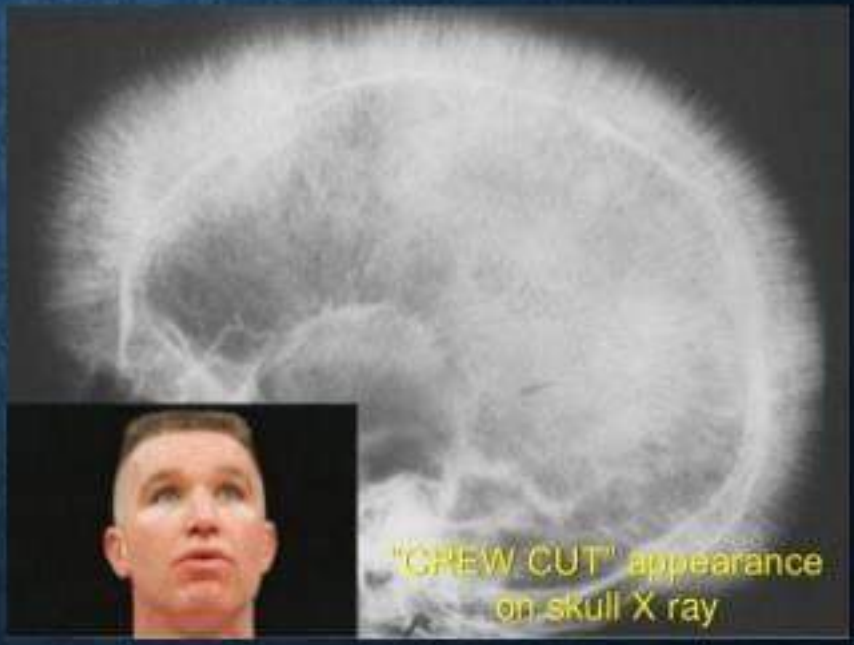
β-Thalassemia facial bone abnormalities. These changes include bossing of the skull; hypertrophy of the maxilla, exposing the upper teeth; depression of nasal bridge; and periorbital puffiness



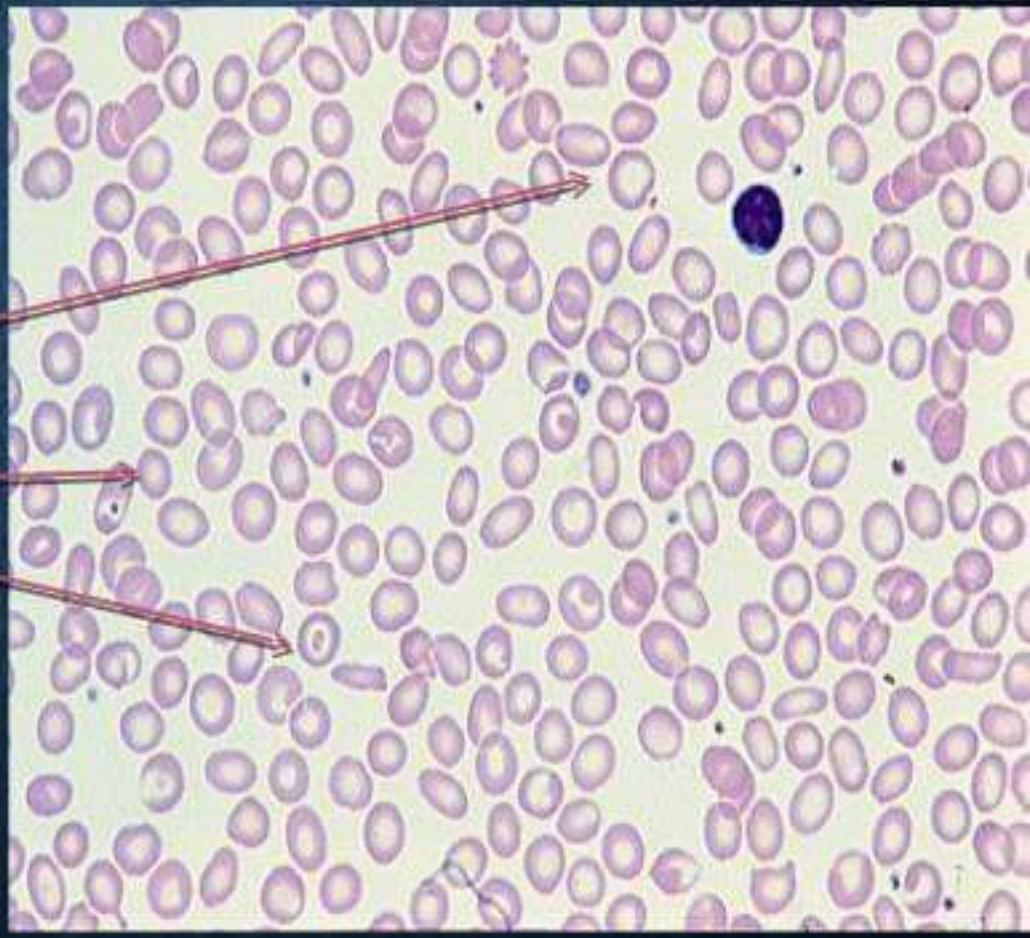
β-Thalassemia major. Note the pallor, short stature, massive hepatosplenomegaly, and wasted limbs in this undertransfused case of β-thalassemia major

BETA THALASSEMIA MAJOR





- Thalassemias
- Smear Characteristics
  - Hypochromia
  - Microcytosis
  - Target Cells
  - Tear Drops



Thalassemia mnemonic by Medicowesome

2014 - THAL

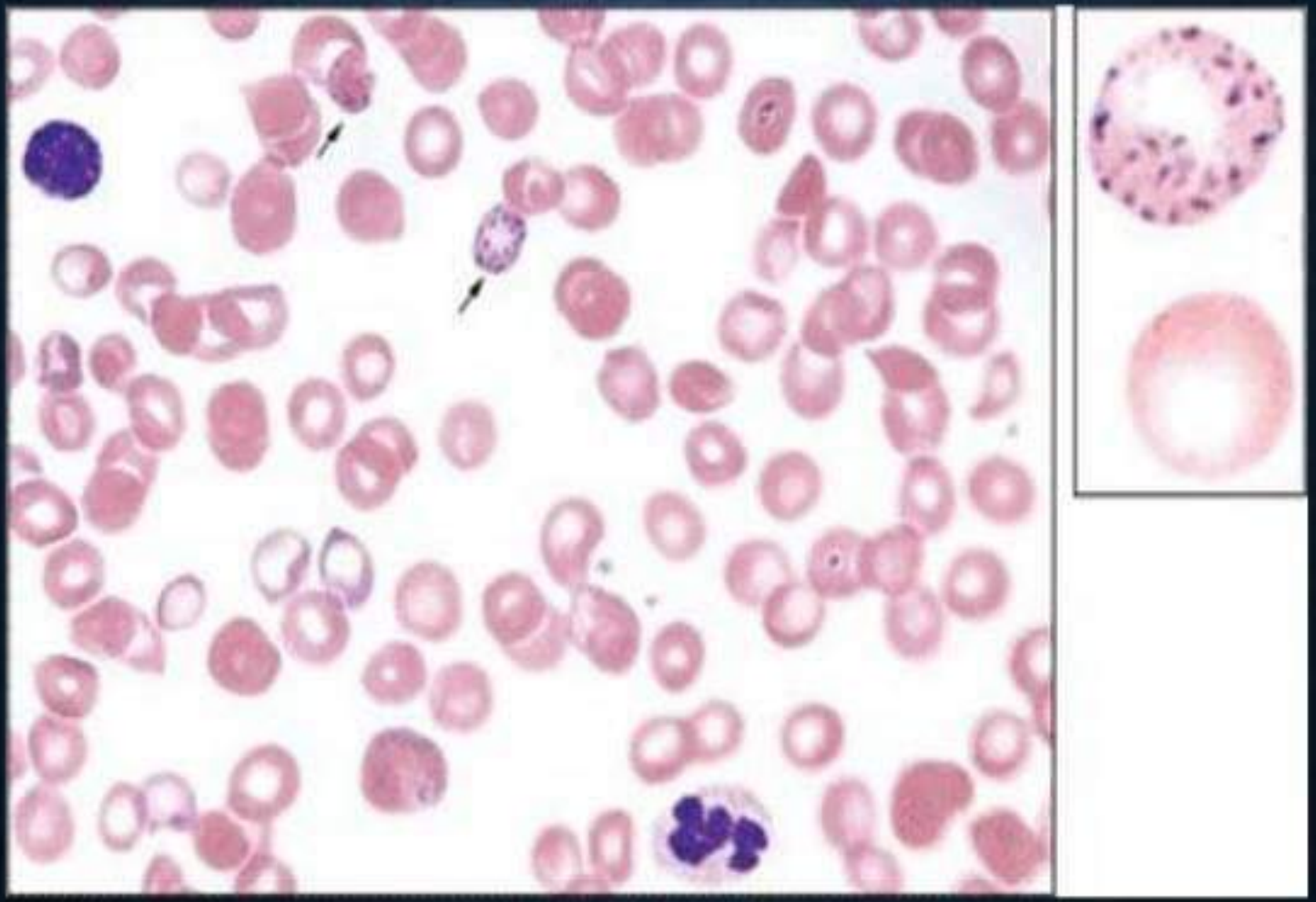
Target cells 

Howell Jolly bodies 

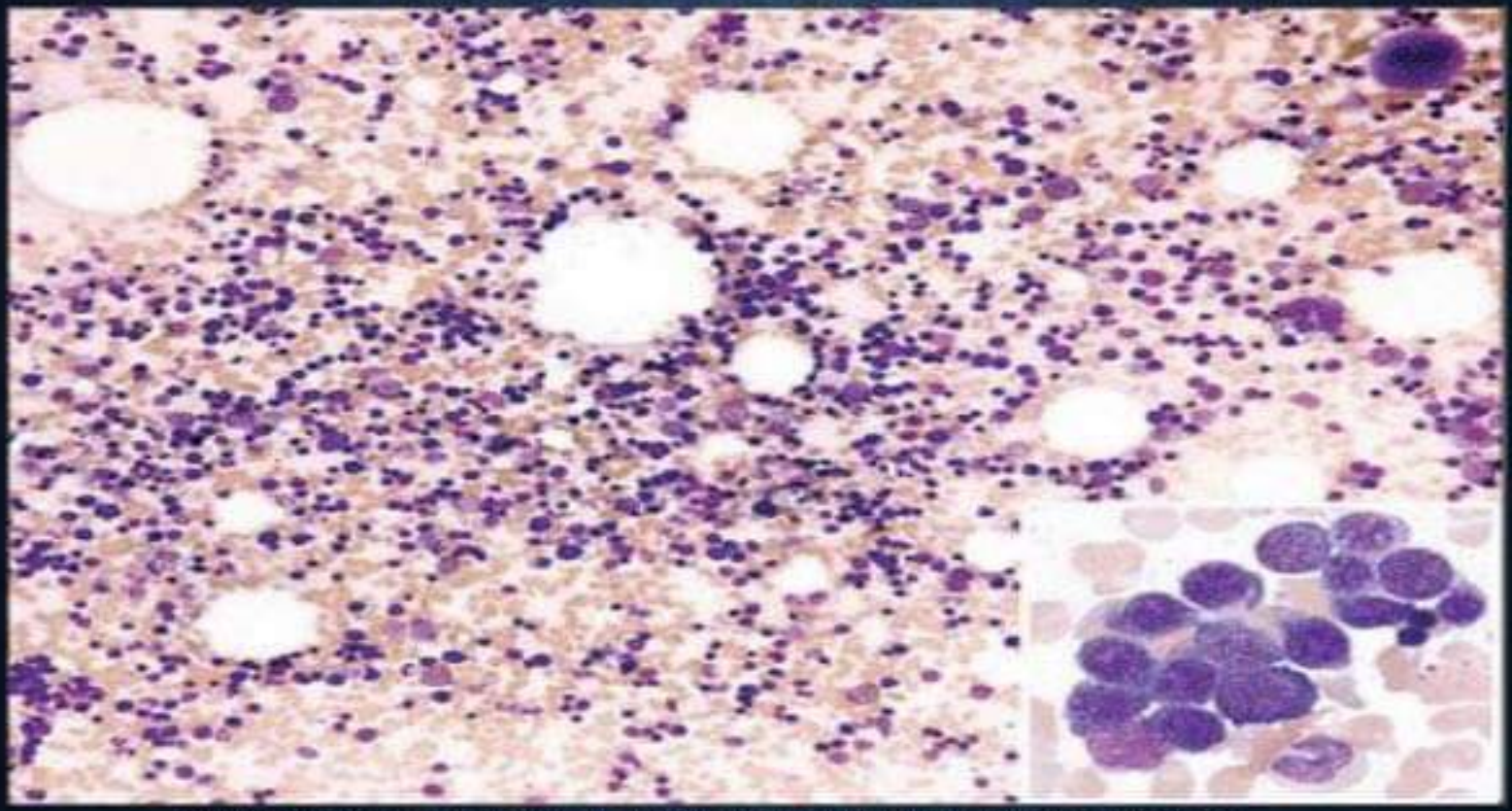
Anisocytosis

Low MCV

(Hypochromic microcytic anemia)

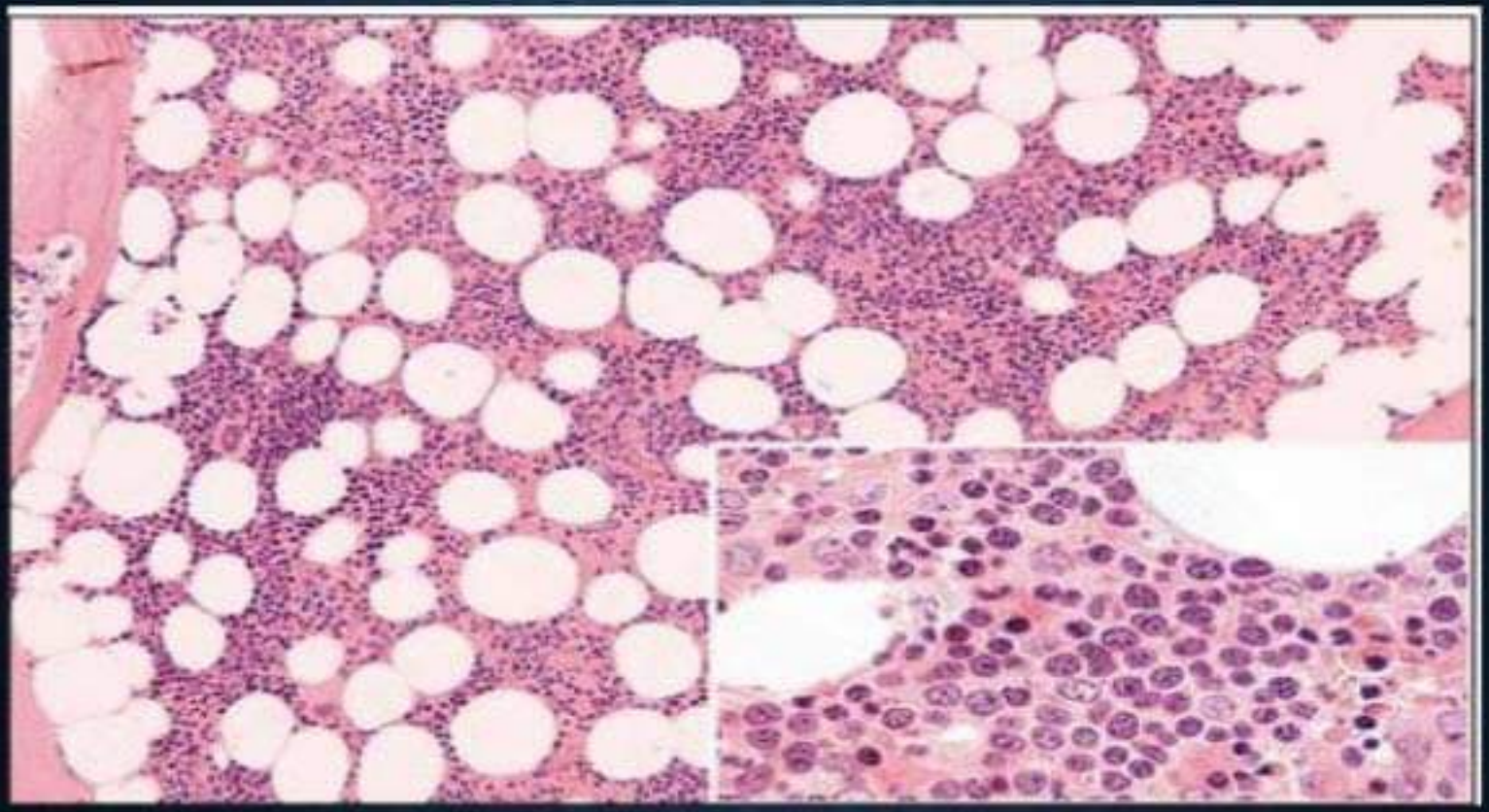


Bone marrow Aspirate



THE BONE MARROW HAS INCREASED NUMBERS OF ERYTHROID PRECURSORS (A LOW MYELOID TO ERYTHROID RATIO) RELATED TO THE INCREASED PERIPHERAL RBC DESTRUCTION IN THIS DISEASE.

Bone marrow Biopsy



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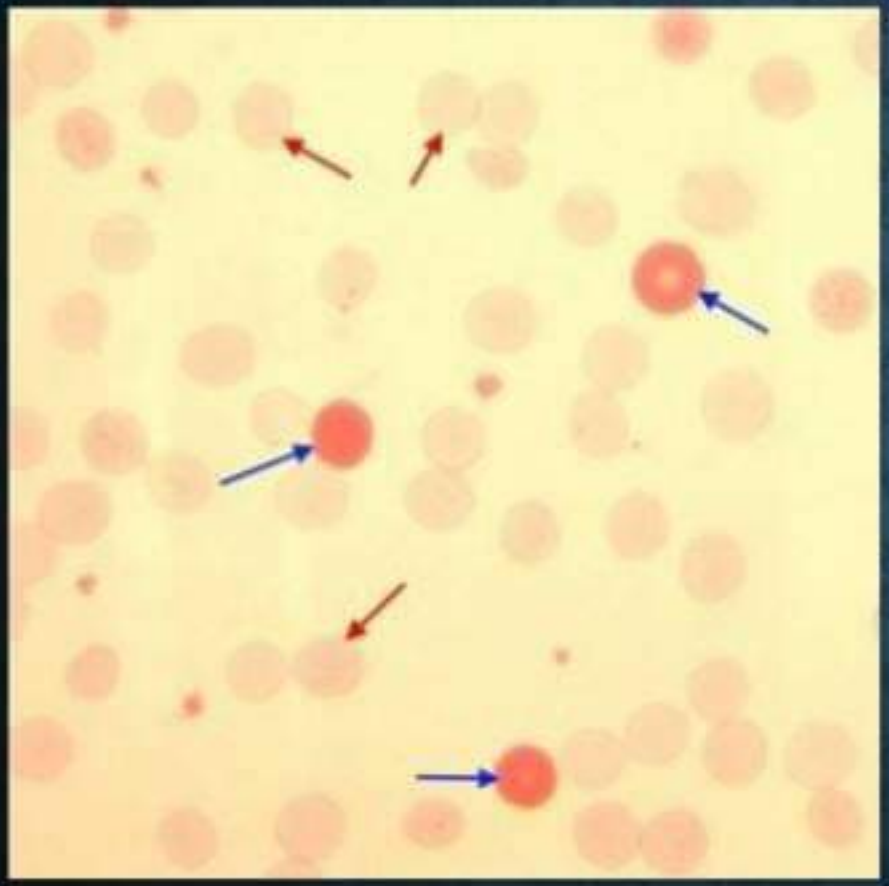
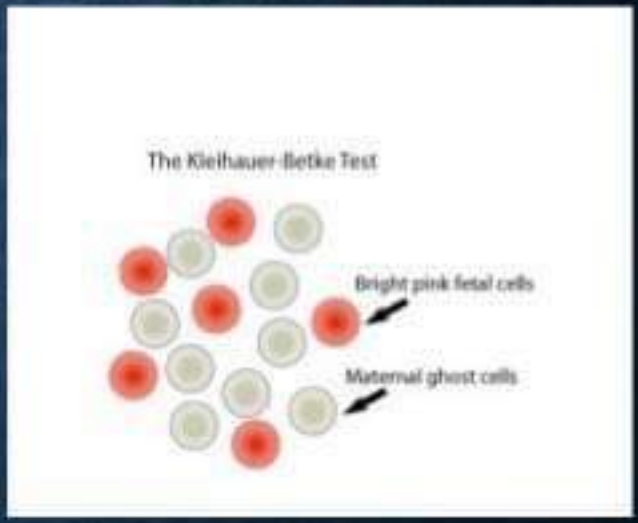
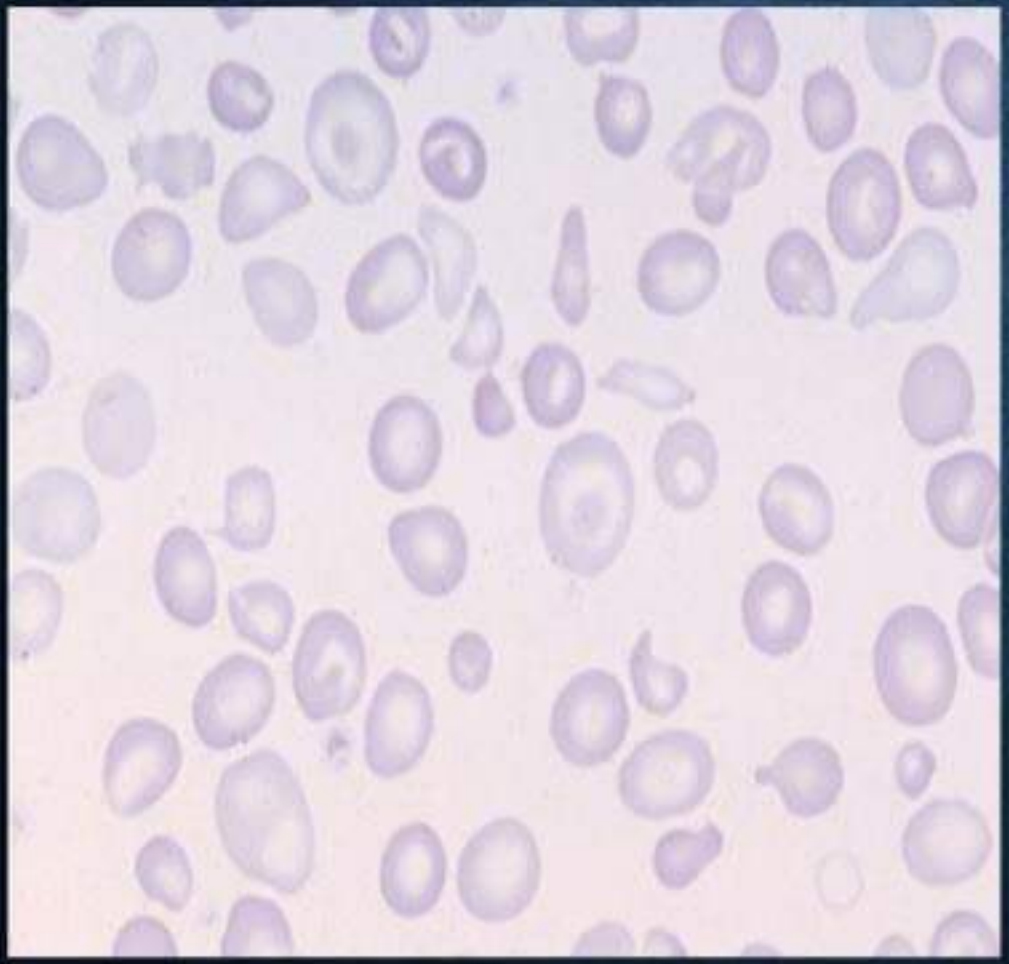


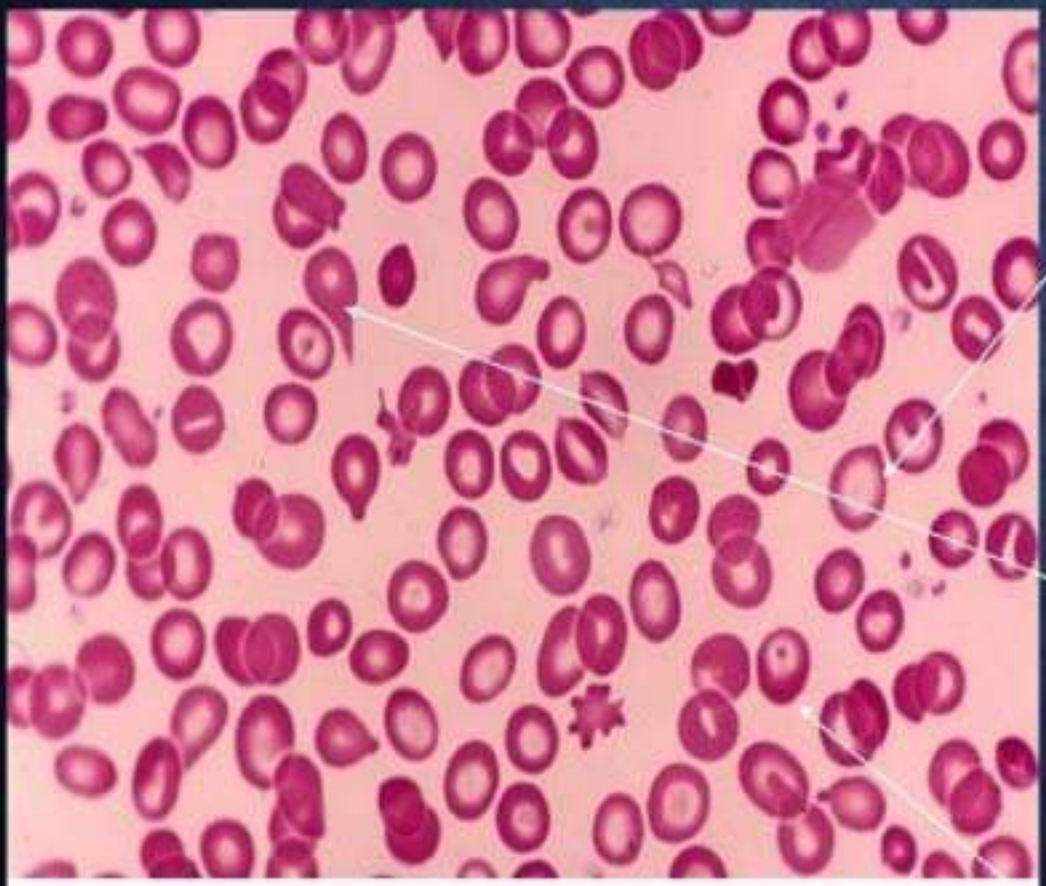


Kleihauer Betke test. This peripheral blood from a postpartum woman with fetomaternal hemorrhage demonstrates HbF containing fetal cells (*dark red) in a background* of maternal cells (ghost-like cells).



MODERATE DEGREE OF ANISOPOIKILOCYTOSIS, MICROCYTIC HYPOCHROMIC, TARGET CELLS,

BETA THALASSEMIA MINOR



MICROCYTOSIS

HYPOCHROMIA

ANISOPOIKILOCYTO-SIS

TEAR DROP CELL

TARGET CELL

Source: Lichtman MA, Shafer MS, Felgar RE, Wang N:
Lichtman's Atlas of Hematology: http://www.accessmedicine.com

MENTZER INDEX(M.I)

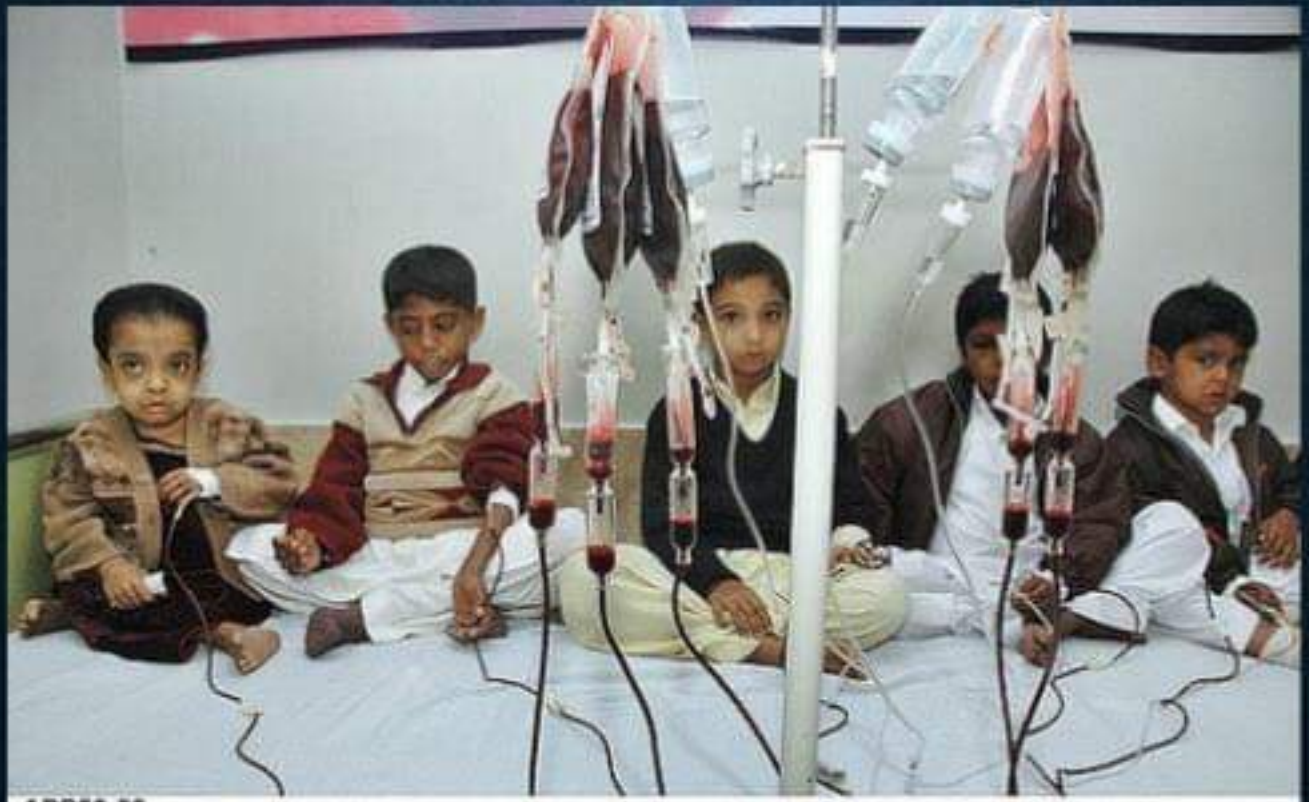
$$M.I = \frac{MCV \quad (fl)}{RED\ CELL\ COUNT\ (millions/ul)}$$

<13 SEEN IN THALASSEMIA

AND

>13 IN IRON DEFICIENCY ANEMIA

When this picture will disappear?



APP83-30
SUKKUR: December 30 – Thalassemia affected children during transfusion of blood at Engro Thalassemia Centre of Sukkur Hospital. APP photo by Jahangir Khan

Physical suffering

In 15 years of life span

Thalassaemia child requires

- 250 uints of Pack cells.
- 4000 Inj of desferroxamine.
- Needle in his body for 40,000 hours of his life.

Iron chelation therapy

Repeated BT causes iron load.
Desferoxamine is Given S.C. for 8 hours every day for 5-6 days a week so as to chelate iron load.