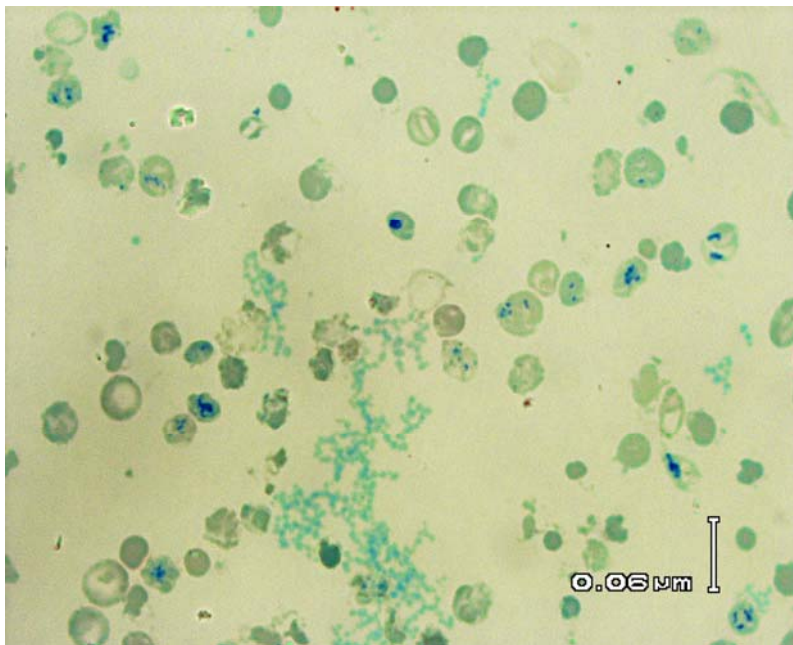
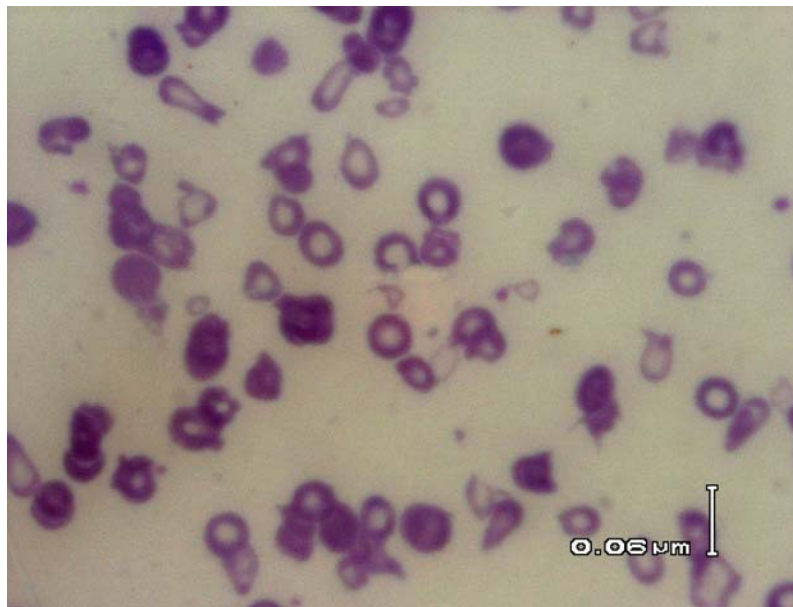
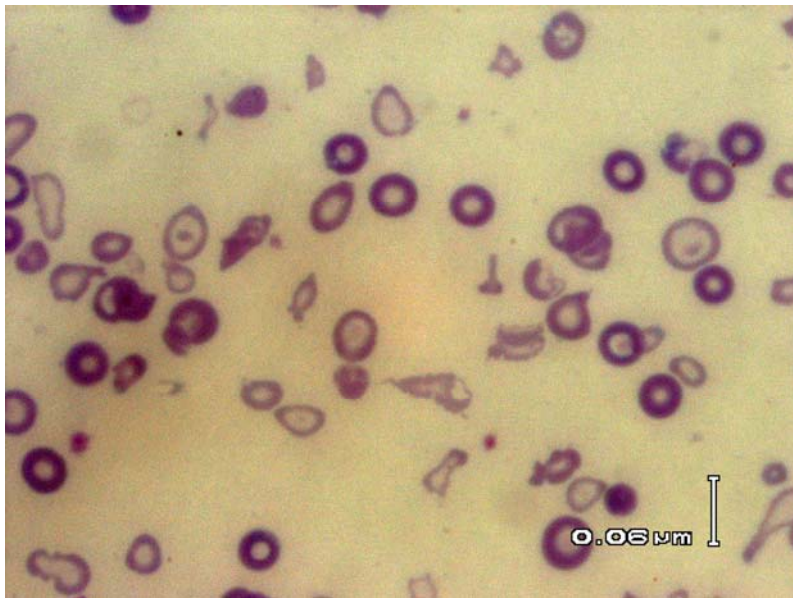




Hb studies

Case 1 (244)

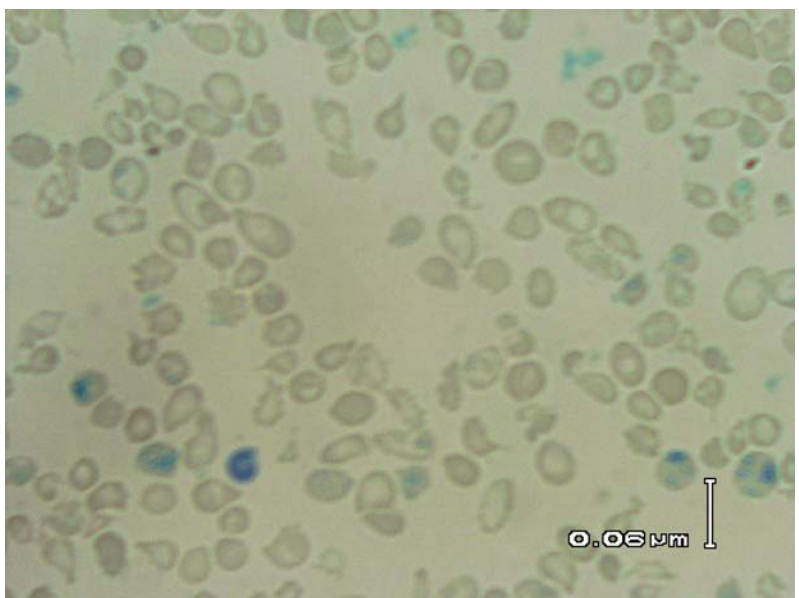
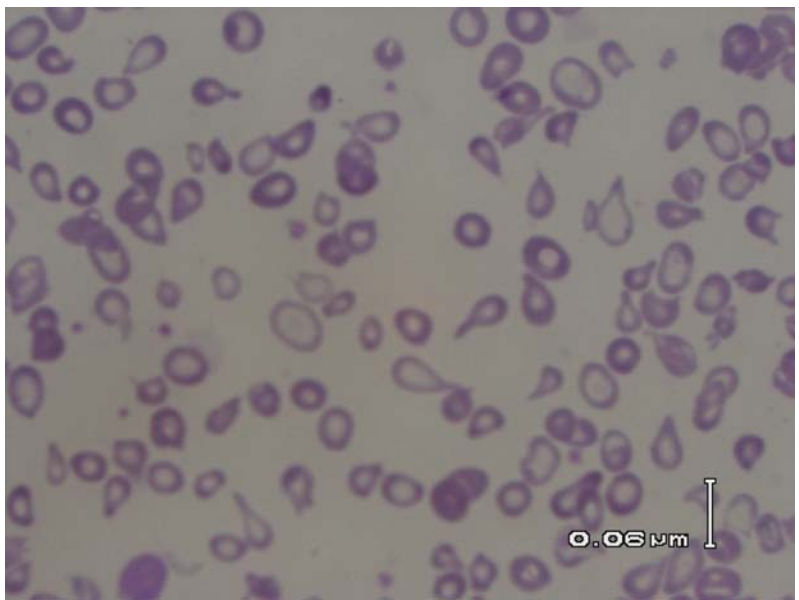
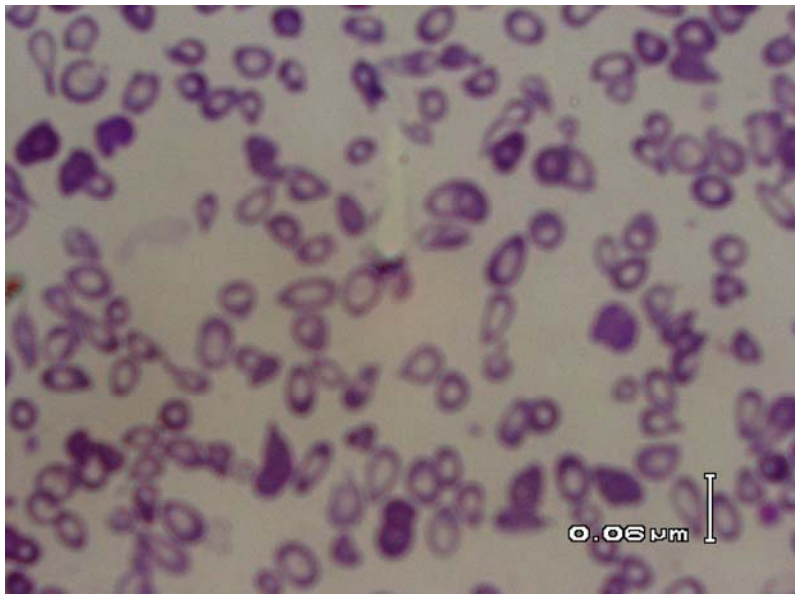
- Female of 36 years age,
- Jutt, Punjabi
- Transfusion dependant for the last 25 years age
- Jaundice for 10 years
- Last transfusion 06 days back
- Liver palpable 7 cms
- spleen palpable 5.5 cms



Hb studies

Case 2 (245)

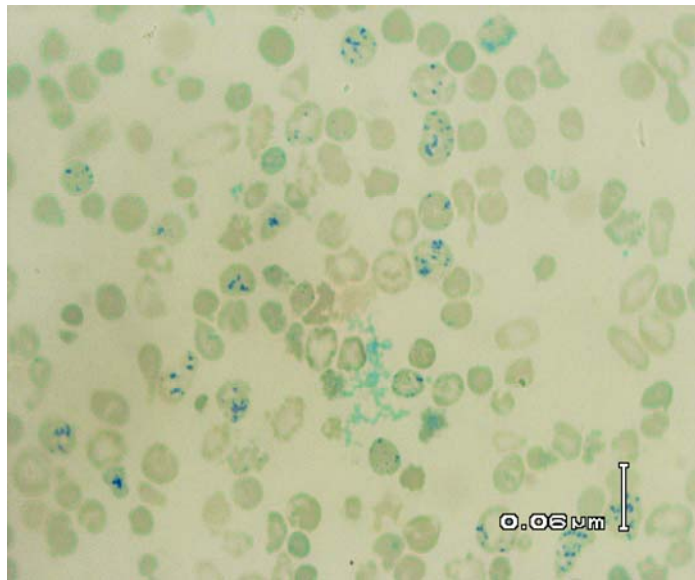
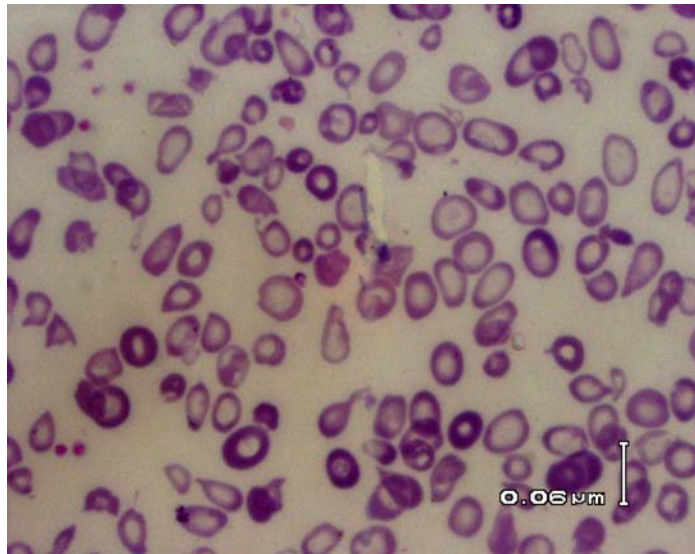
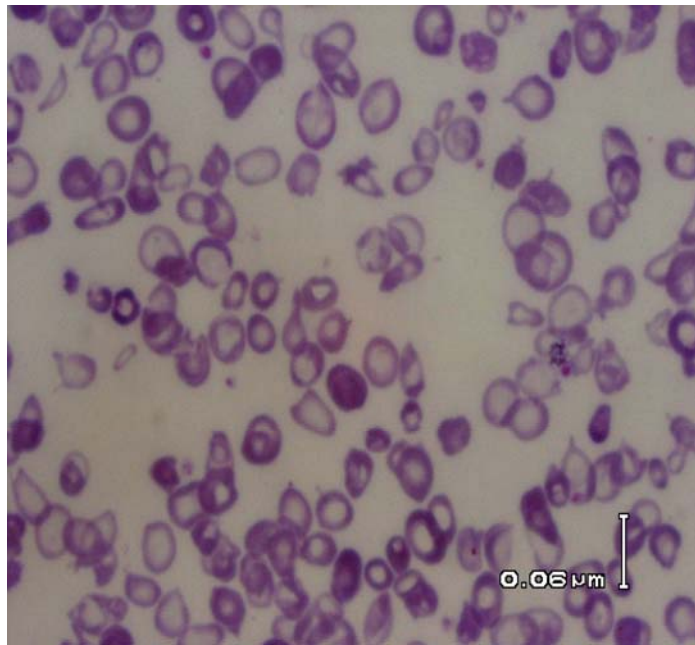
- (sister of 244)
- Female of 26 years age,
- Jutt, Punjabi
- Progressive pallor
- No blood transfusions
- Liver edge palpable
- spleen palpable 6 cms



Hb studies

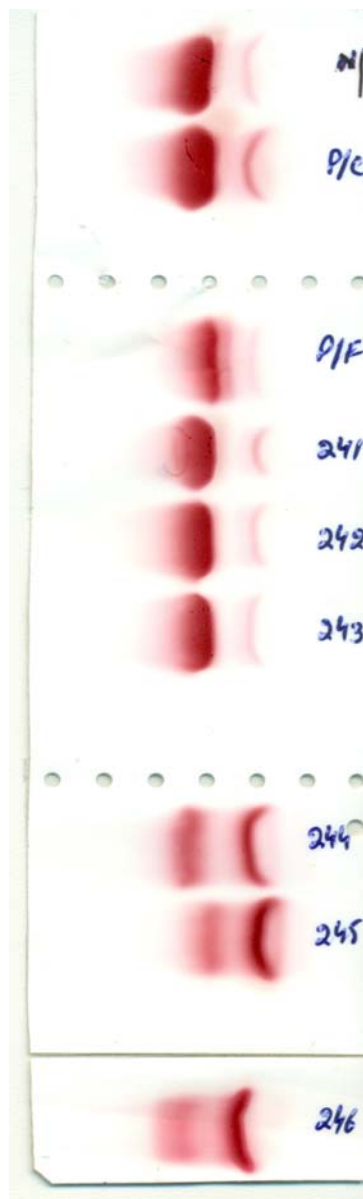
Case 3 (246)

- (sister of 244)
- Female of 22 years age
- Jutt Punjabi
- Progressive pallor
- fever off and on
- Transfused 12 years back
- Liver edge palpable
- spleen palpable 5 cms

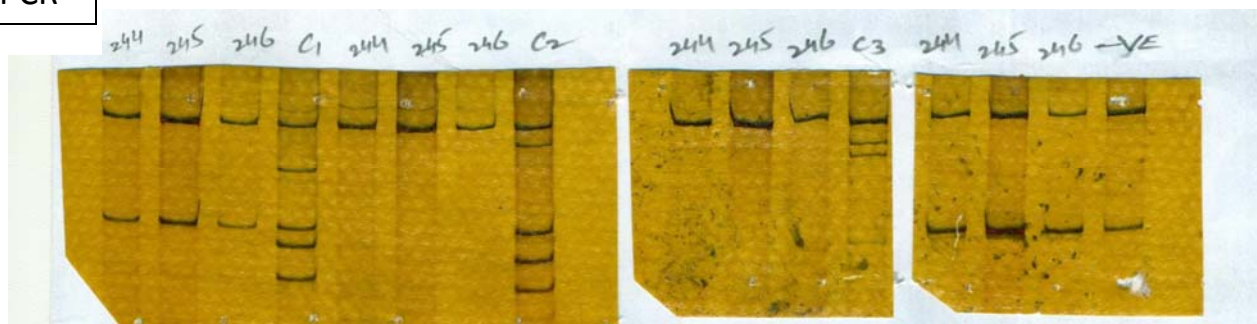


Parameter		Patient 1(244)	Patient 2(245)	Patient 3(246)
RCC	x10 ¹² /L	1.52	3.23	3.37
Hb	g/dl	3.2	5.7	5.8
MCV	fl	68.4	60.7	61.7
MCH	pg	21.1	17.6	17.5
MCHC	g/dl	30.8	29.2	28.3
RDW				
Retics	%	6.4	4.0	5.0
N RBCs	/100 wbc	1	15	0
TLC	x10 ⁹ /L	3.44	10.6	6.92
Platelets	x10 ⁹ /L	146	177	112

Hb Electrophoresis on Cellulose Acetate at pH 8.6



PCR



Opinion

- Case 1-3(244, 245, 246): Compound heterozygotes for IVS1-5 (Beta Thalassaemia) and HbE

Compound heterozygotes for Beta thalassaemia and HbE

- High frequency in South-East Asia
- Has been reported from different parts of India
- The clinical picture is heterogeneous
- In case presentation with severe anemia and early age of onset the disease is similar to homozygous Beta-thalassemia
- In case presentation with mild anemia and later age of onset occasional need for blood transfusion or asymptomatic
- HbE is asymptomatic in homozygous state.
- The severity of phenotype is, therefore, dependent
- on the type of Beta thal mutation
- HbE and HbF levels and number of alpha globin genes which tend to reduce the severity of disease by altering the ratio of imbalance of alpha and globin chains.
- One of the common thalassemia mutation is IVS-1,5 (G->C) in India.