



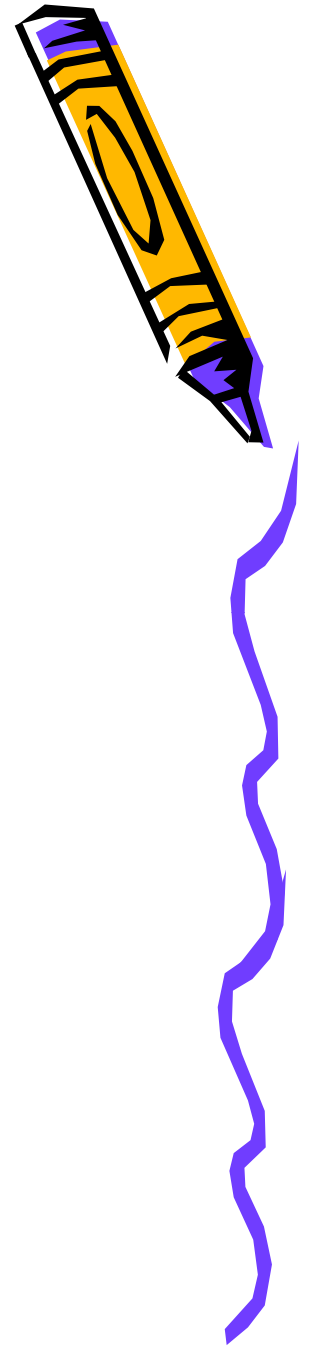
Thalassaemia: Usual to the most unusual

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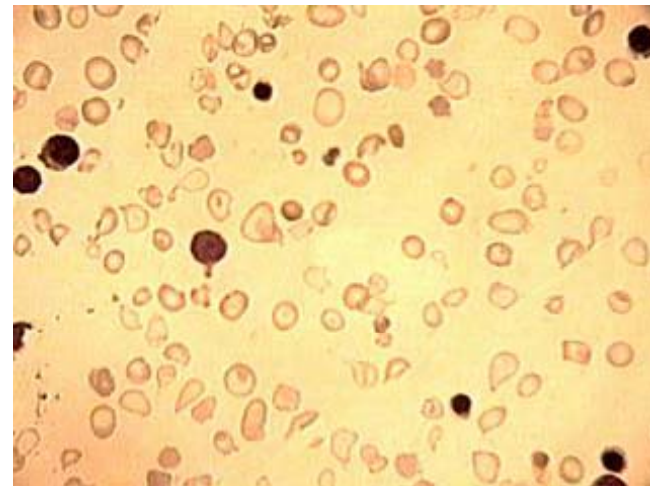
Armed Forces Institute of Pathology



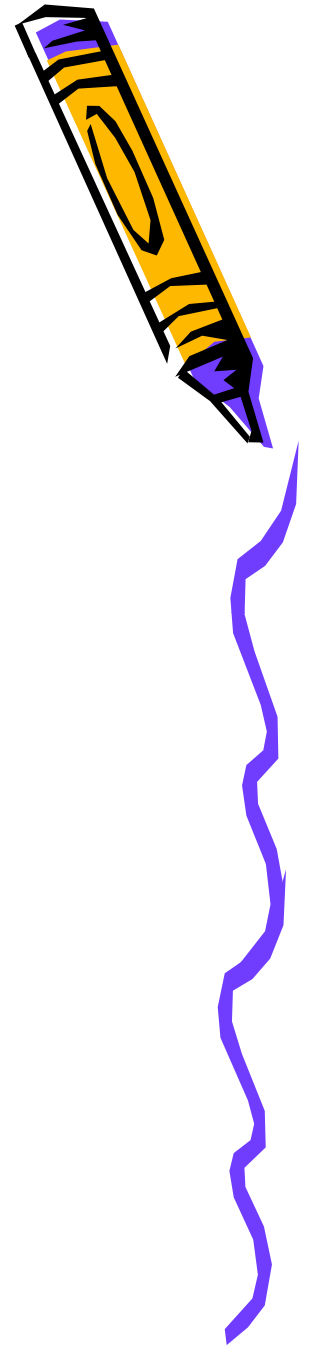
- Age: 6 months
- Sex: Female
- Ethnic: Punjabi
- Consanguinity: 1st cousins
- Complaints:
 - Progressive pallor 3 months
 - Fever off and on 1 month
- Transfusions: Nil
- Examination:
 - Liver: Nil
 - Spleen: 3 cm



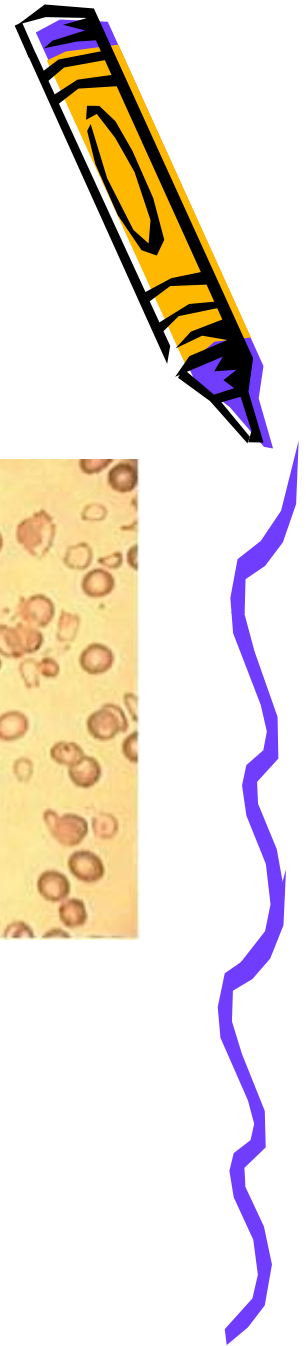
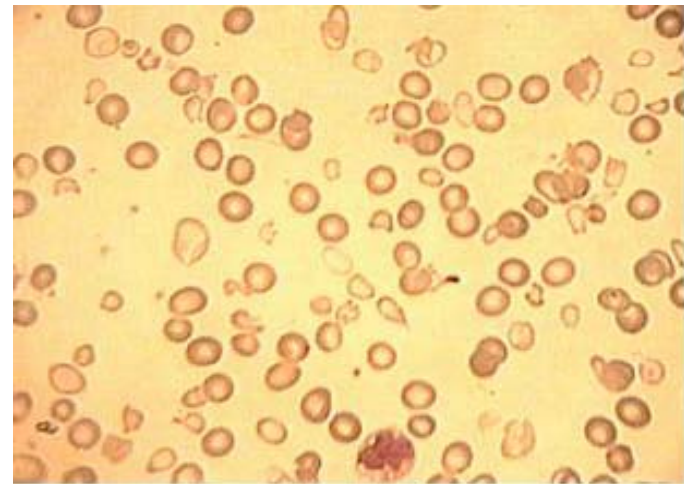
- WBC: 23.3 X 10⁹/L
- RBCs: 3.52 X 10¹²/L
- Hb: 7.0 g/dl
- MCV: 67.5 fl
- MCV: 19.2 pg
- Platelets: 533
- Reticulocytes: 0.2%
- Hb-F: 97.1%
- Hb-A2: 2.9%
- Electrophoresis: Hb F and A2



- Age: 2 years
- Sex: Female
- Ethnic: Punjabi
- Consanguinity: 1st cousins
- Complaints:
 - Progressive pallor since 3 months of age
 - Fever off and on 1 month
- Transfusions: Multiple
- Examination:
 - Liver: 2 cm
 - Spleen: 4 cm



- WBC: $7.3 \times 10^9/L$
- RBCs: $3.12 \times 10^{12}/L$
- Hb: 8.2 g/dl
- MCV: 77.5 fl
- MCV: 22.2 pg
- Platelets: 233
- Reticulocytes: 0.2%
- Hb-F: 7.1%
- Hb-A2: 2.3%
- Electrophoresis: Hb A, F & A2





Father

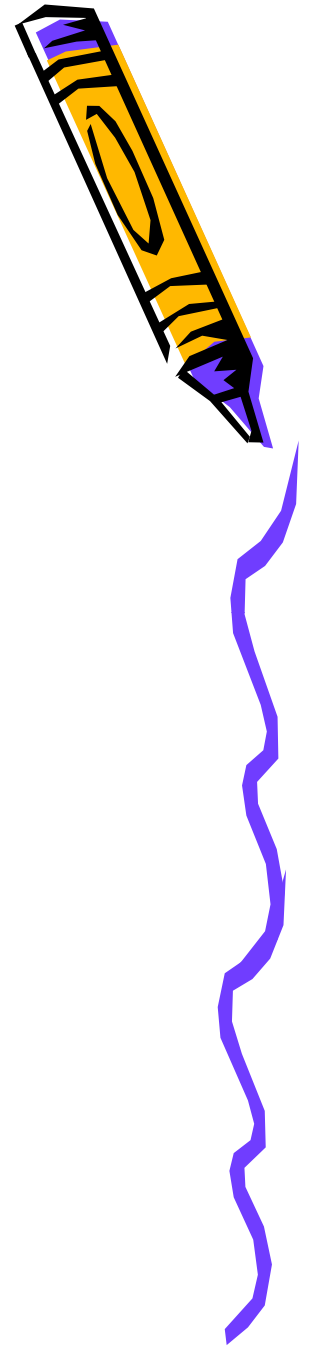
- WBC: $7.3 \times 10^9/L$
- RBCs: $7.12 \times 10^{12}/L$
- Hb: 14.8 g/dl
- MCV: 69.1 fl
- MCV: 20.8 pg
- Platelets: 195

Mother

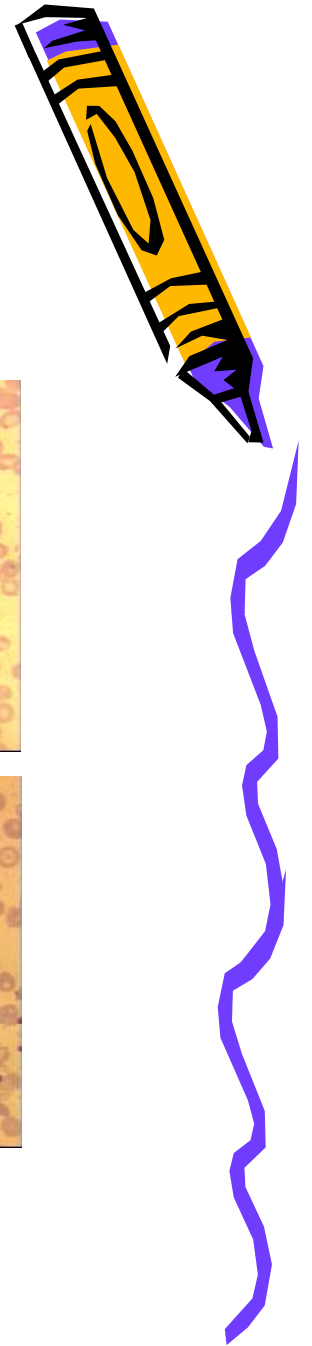
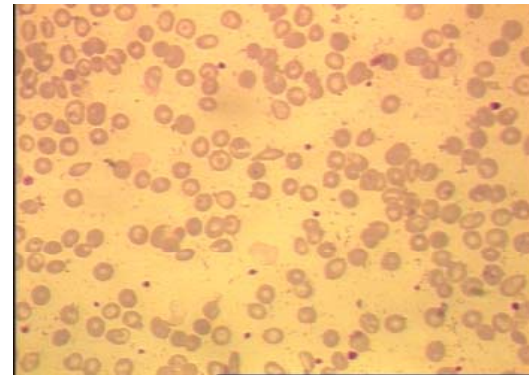
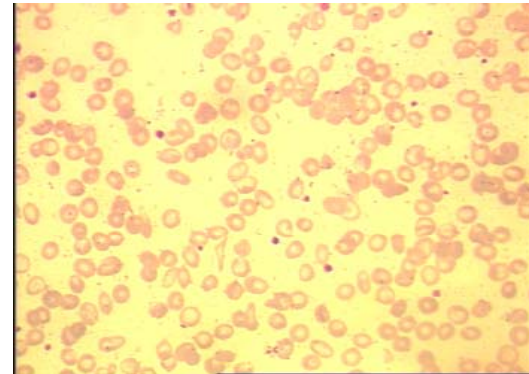
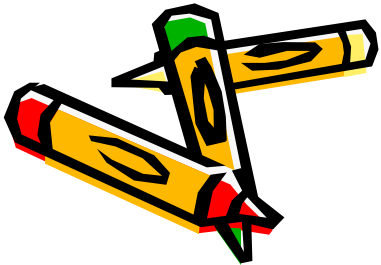
- WBC: $6.8 \times 10^9/L$
- RBCs: $5.48 \times 10^{12}/L$
- Hb: 10.6 g/dl
- MCV: 64.5 fl
- MCV: 19.2 pg
- Platelets: 270



- Age: 16 years
- Sex: Male
- Ethnic: Pathan (Bhitani)
- Consanguinity: 1st cousins
- Complaints:
 - Progressive pallor 6 years
 - Ill health 2 years
- Transfusions: Ten times
- Examination:
 - Liver: 2 cm
 - Spleen: 8 cm



- WBC: $7.23 \times 10^9/L$
- RBCs: $3.37 \times 10^{12}/L$
- Hb: 7.4 g/dl
- MCV: 70.0 fl
- MCV: 22.2 pg
- Platelets: 198
- Reticulocytes: 3.5%
- Hb-F: 15.3%
- Hb-A2: 4.6%

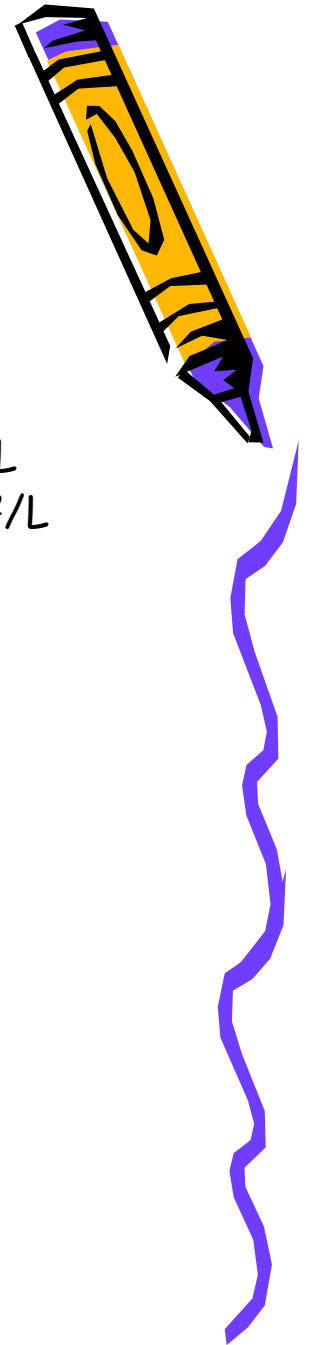
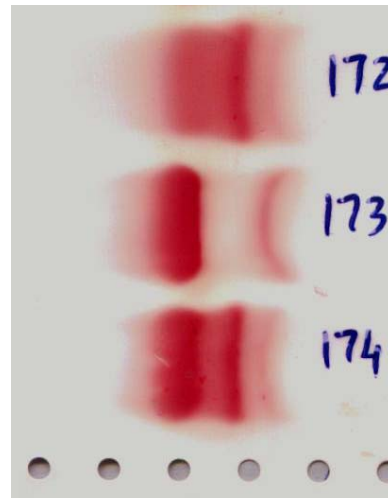


Father (173)

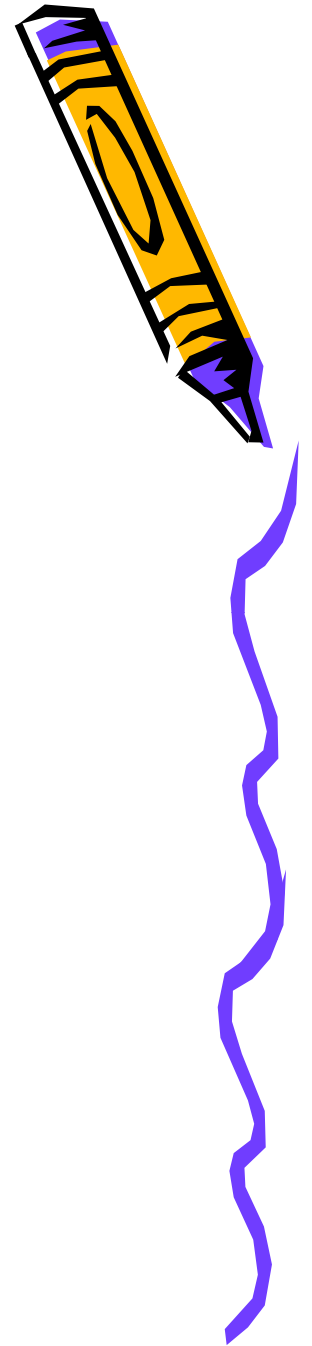
- WBC: $7.3 \times 10^9/L$
- RBCs: $6.12 \times 10^{12}/L$
- Hb: 13.3 g/dl
- MCV: 69.1 fl
- MCV: 20.8 pg
- Platelets: 295

Mother (174)

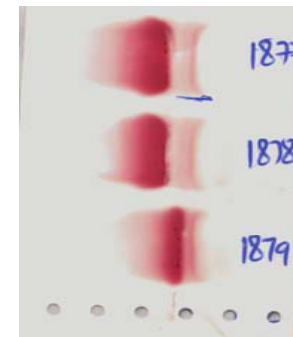
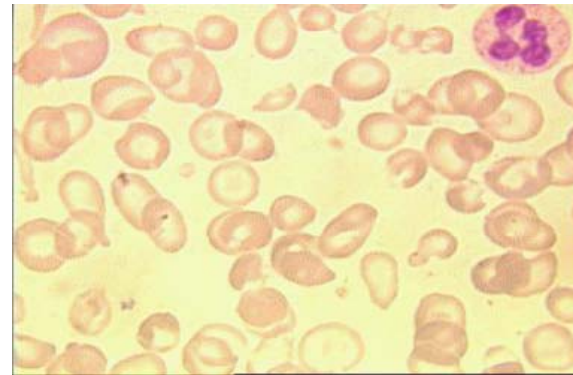
- WBC: $6.8 \times 10^9/L$
- RBCs: $4.18 \times 10^{12}/L$
- Hb: 10.6 g/dl
- MCV: 84.5 fl
- MCV: 27.2 pg
- Platelets: 270



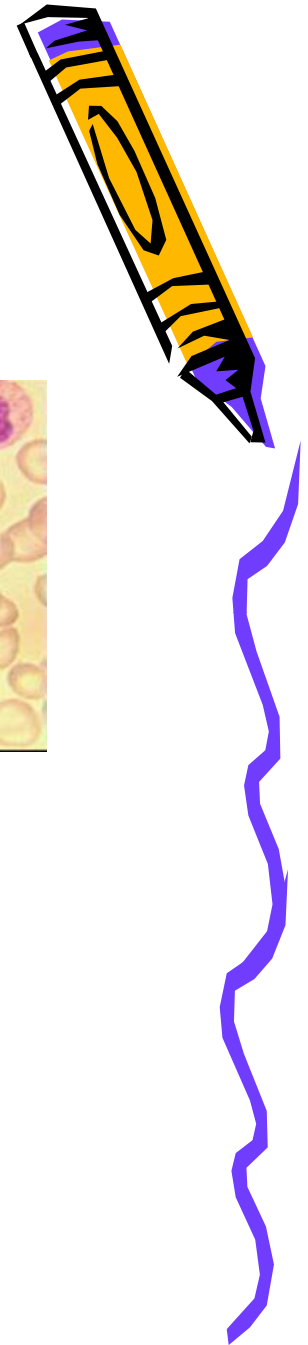
- Age: 17 years
- Sex: Female
- Ethnic: Punjabi
- Consanguinity: Unrelated
- Complaints:
 - Incidental discovery
- Transfusions: Nil
- Examination:
 - Liver: NP
 - Spleen: NP



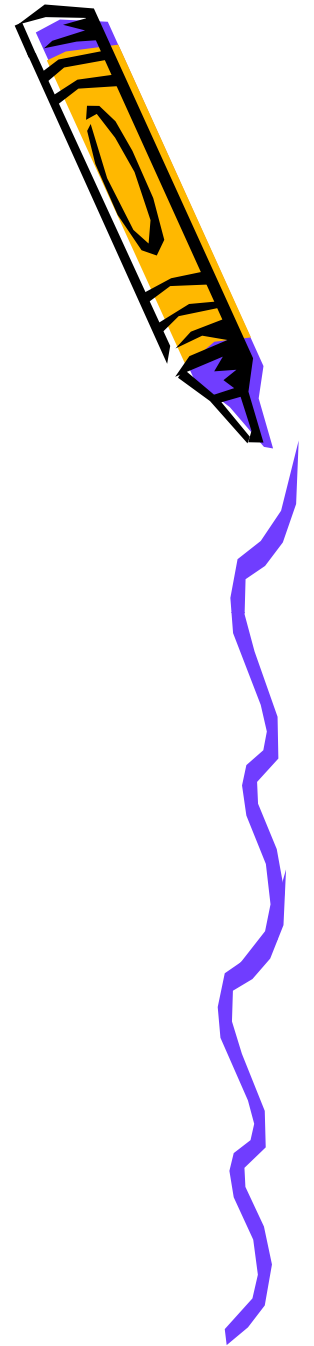
- WBC: $7.73 \times 10^9/L$
- RBCs: $5.21 \times 10^{12}/L$
- Hb: 9.4 g/dl
- MCV: 56.5 fl
- MCV: 18.2 pg
- Platelets: 241
- Reticulocytes: 2.6%



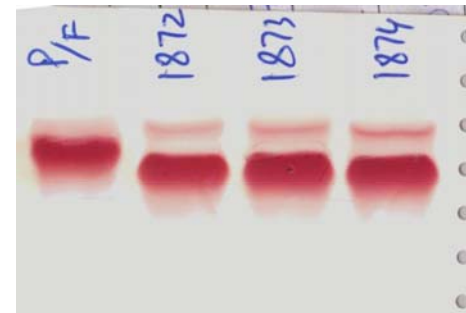
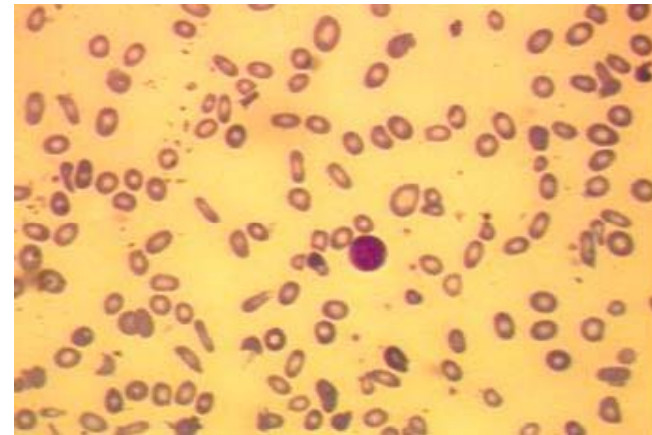
Hb-A2: 4.5%
PCR: Hb-D/Fr 8-9



- Age: 47 years
- Sex: Female
- Ethnic: Punjabi
- Consanguinity: 1st cousins
- Complaints:
 - Weakness: 2 months
- Transfusions: None
- Examination:
 - Liver: NP
 - Spleen: NP



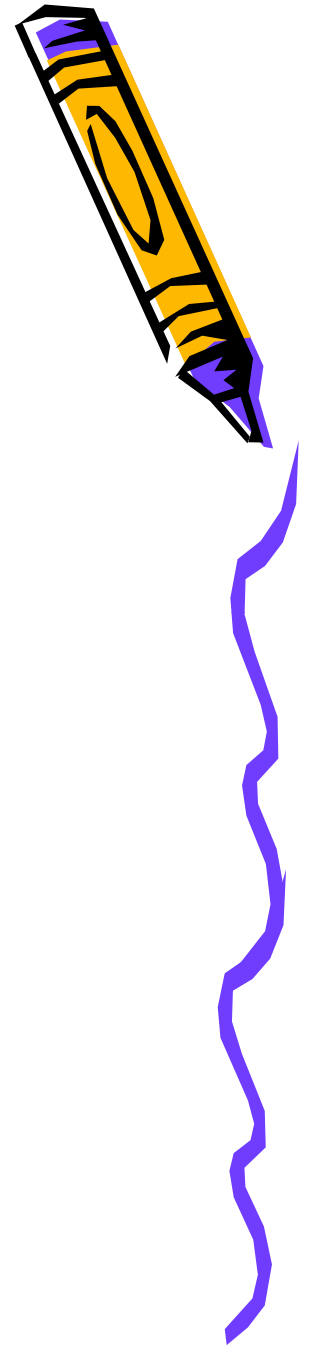
- WBC: 4.75 X 10⁹/L
- RBCs: 2.29 X 10¹²/L
- Hb: 5.2 g/dl
- MCV: 73.8 fl
- MCV: 22.7 pg
- Platelets: 157
- Reticulocytes: 3.0%



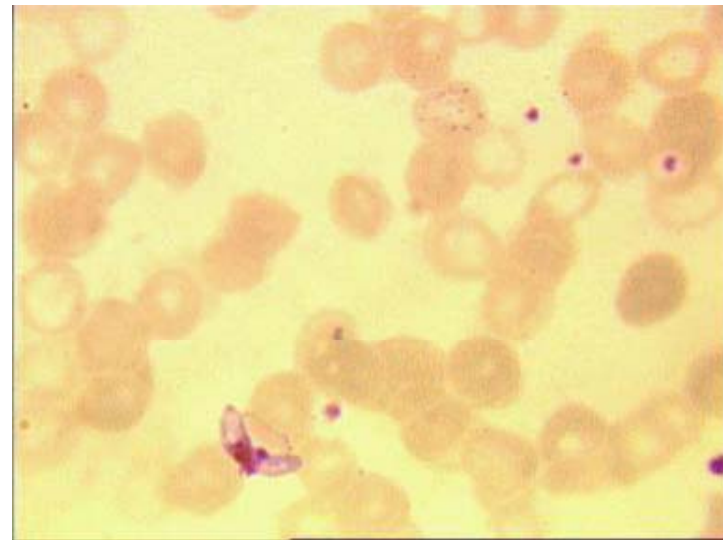
Hb-A2: 5.3%
PCR: Het Cd5



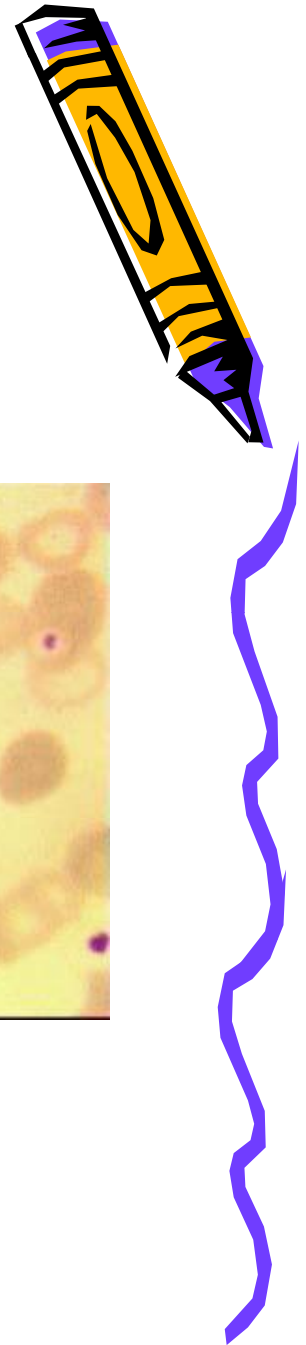
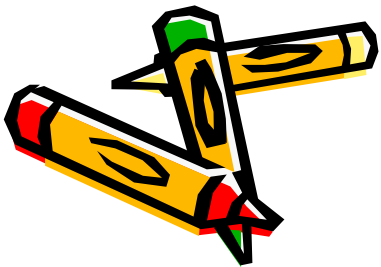
- Age: 26 years
- Sex: Male
- Ethnic: Pathan
- Consanguinity: 1st cousins
- Complaints:
 - Fever with chills 1 week
- Transfusions: Nil
- Examination:
 - Liver: NP
 - Spleen: 2 cm



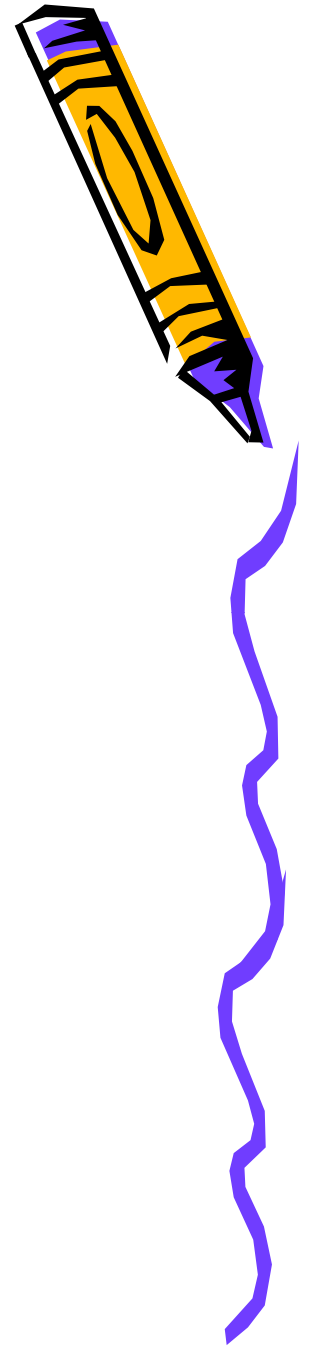
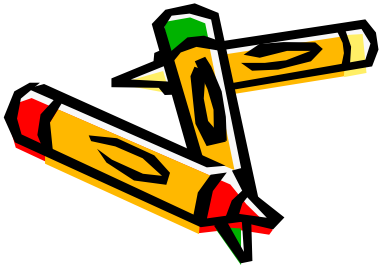
- WBC: 13.23 X 10⁹/L
- RBCs: 4.21 X 10¹²/L
- Hb: 10.5 g/dl
- MCV: 61.5 fl
- MCV: 19.3 pg
- Platelets: 171
- Reticulocytes: 4.2%



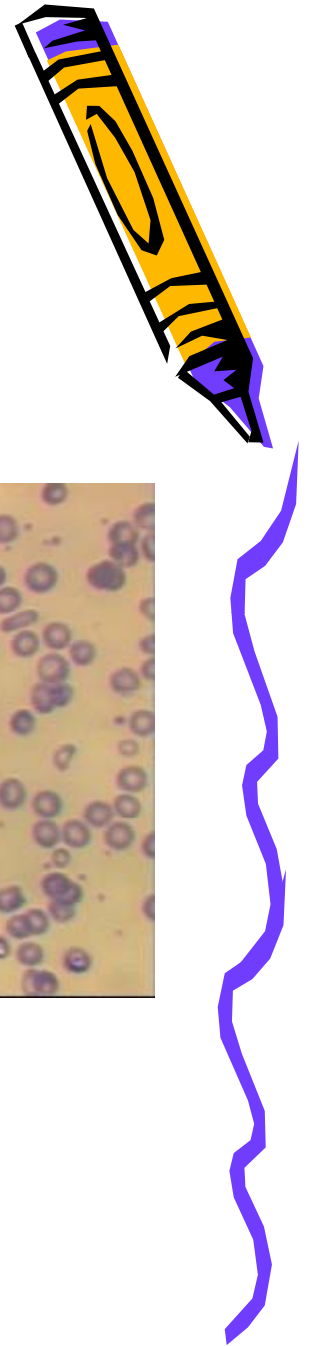
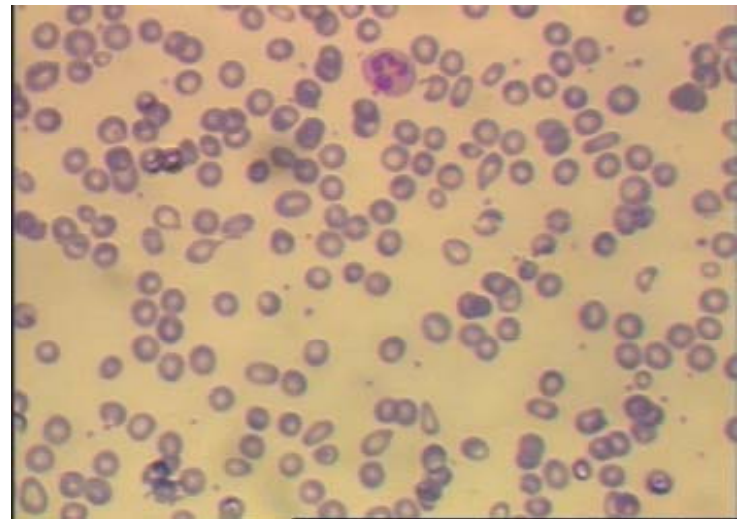
Hb-A2: 5.1%
PCR: Fr 8-9

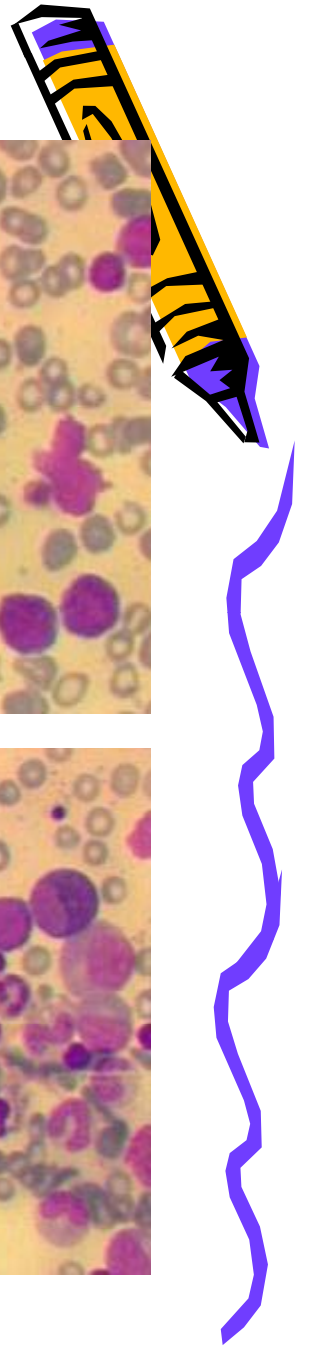
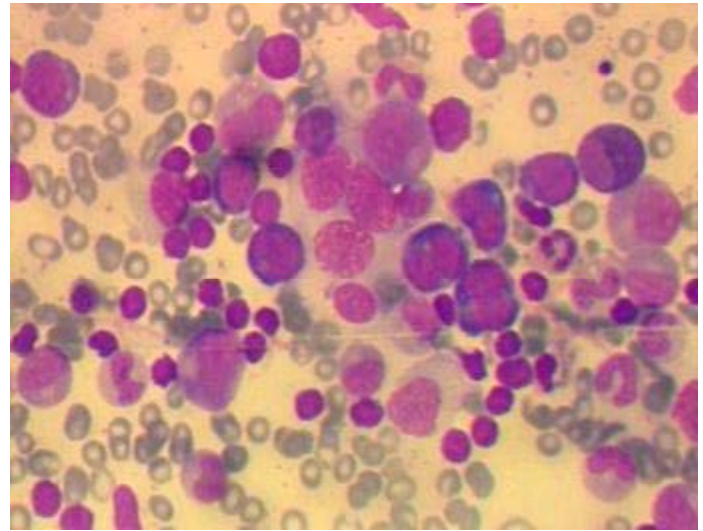
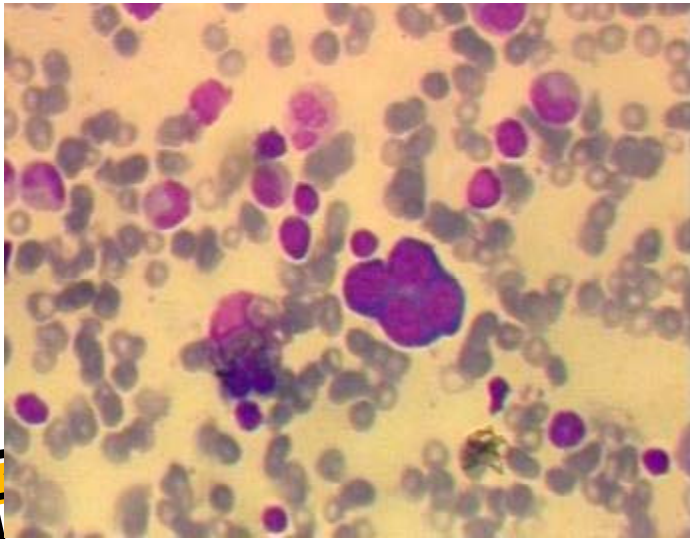
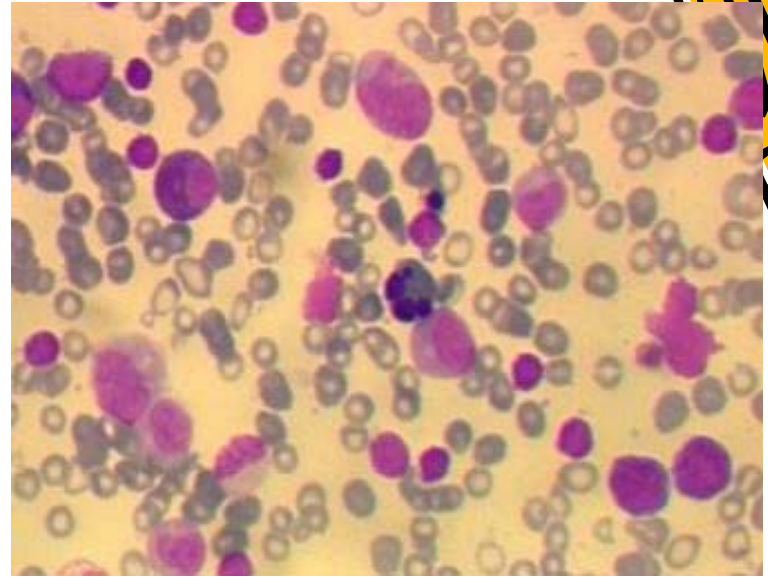
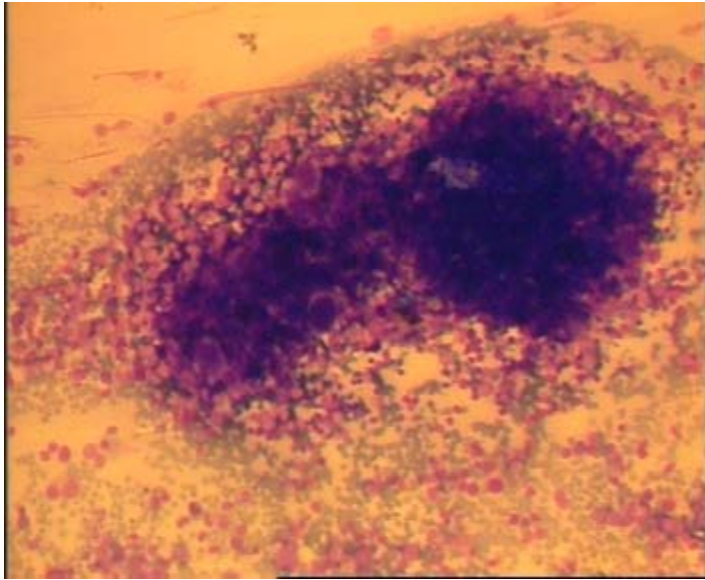


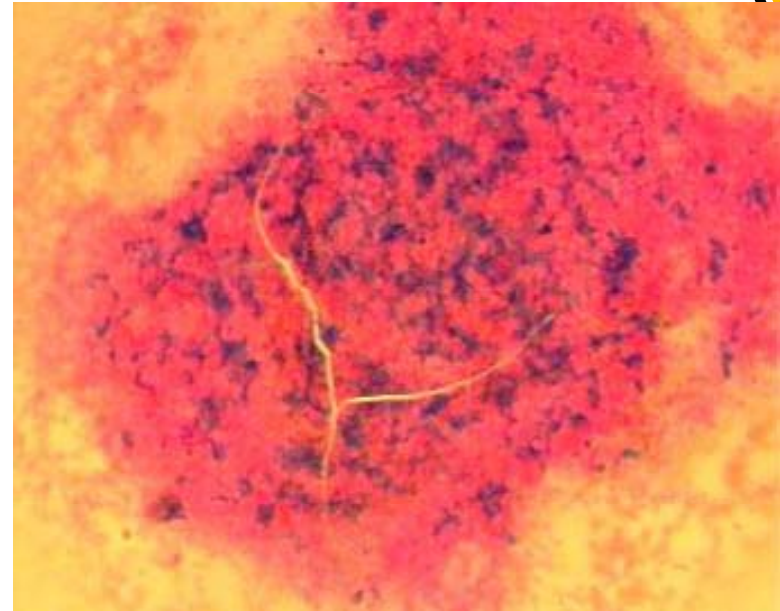
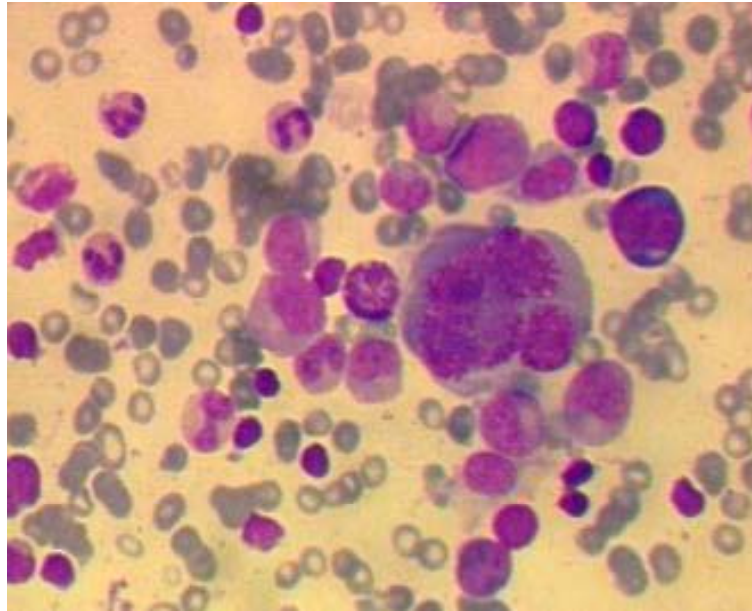
- Age: 1 year
- Sex: Male
- Ethnic: Punjabi
- Consanguinity: 1st cousins
- Complaints:
 - Progressive pallor 6 months
 - Fever off and on 6 months
- Transfusions: Thrice
- Examination:
 - Liver: Nil
 - Spleen: tip



- WBC: $13.23 \times 10^9/L$
- RBCs: $3.21 \times 10^{12}/L$
- Hb: 8.4 g/dl
- MCV: 78.5 fl
- MCV: 26.2 pg
- Platelets: 341
- Reticulocytes: 0.2%
- Hb-F: 0.9%
- Hb-A2: 2.9%
- Electrophoresis: No abnormal band seen







PCR: Heterozygous del 619



