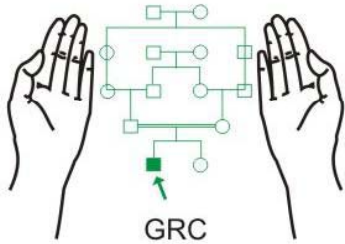




# Difficult Cases of Thalassaemia

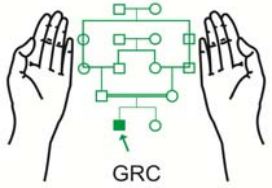
Maj Gen (R) Suhaib Ahmed, HI (M)  
MBBS; MCPS; FCPS (Pak); PhD (London)



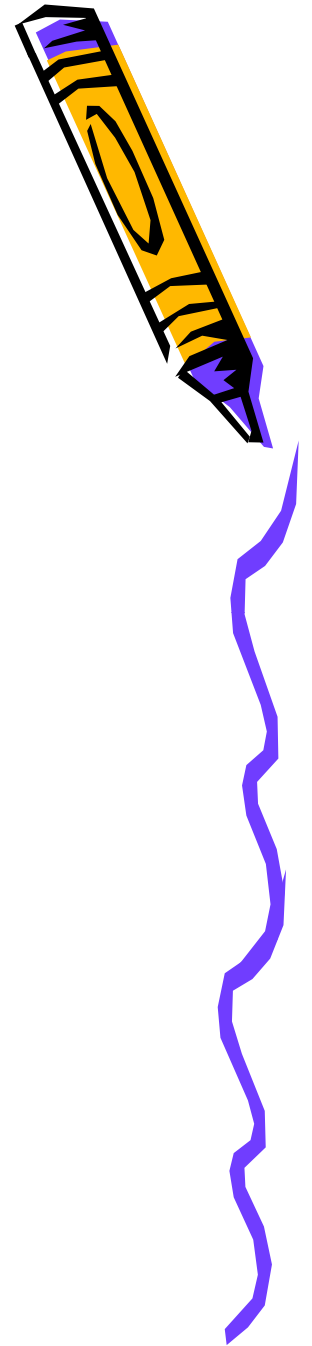
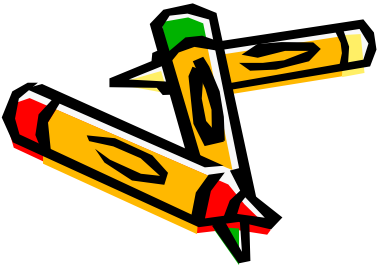
**Genetics Resource Centre (GRC)**

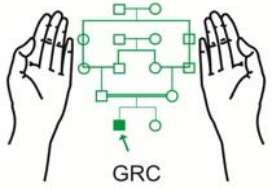


[www.grcpk.com](http://www.grcpk.com)

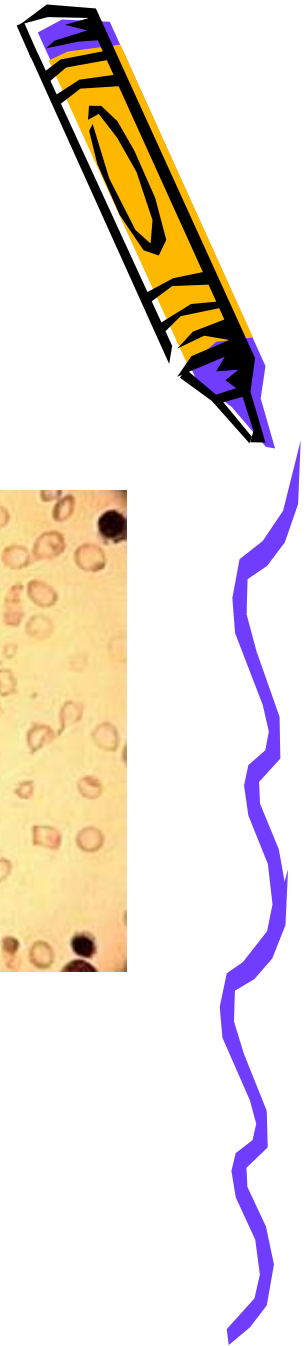
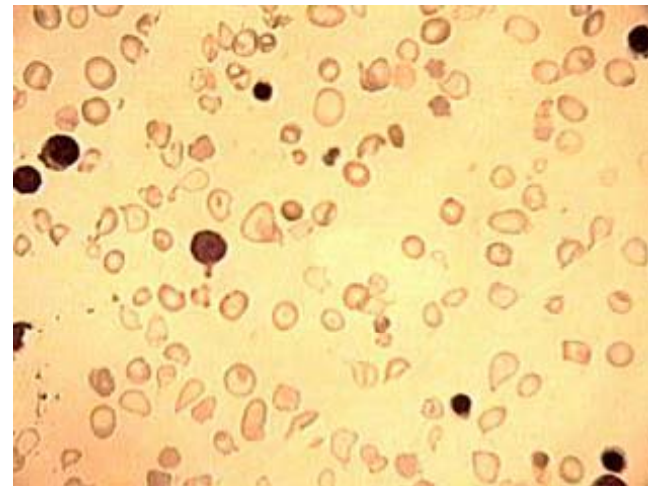


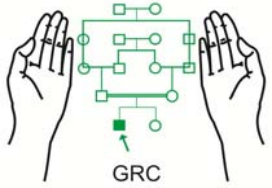
- Age: 6 months
- Sex: Female
- Ethnic: Punjabi
- Consanguinity: 1st cousins
- Complaints:
  - Progressive pallor 3 months
  - Fever off and on 1 month
- Transfusions: Nil
- Examination:
  - Liver: Nil
  - Spleen: 3 cm



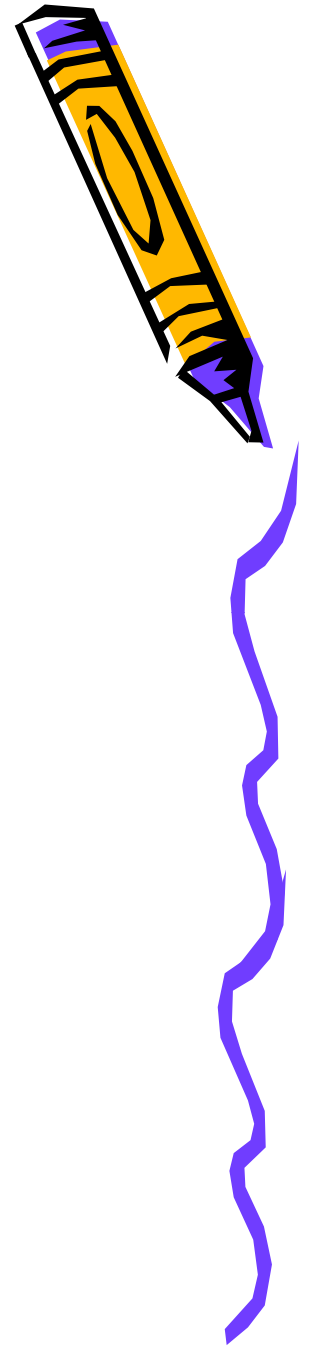
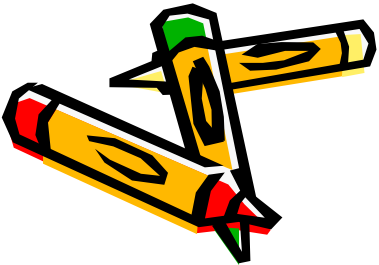


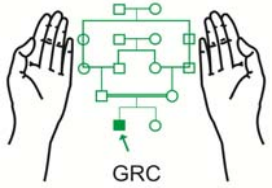
- WBC:  $23.3 \times 10^9/L$
- RBCs:  $3.52 \times 10^{12}/L$
- Hb: 7.0 g/dl
- MCV: 67.5 fl
- MCV: 19.2 pg
- Platelets: 533
- Reticulocytes: 0.2%
- Hb-F: 97.1%
- Hb-A2: 2.9%
- Electrophoresis: Hb F and A2



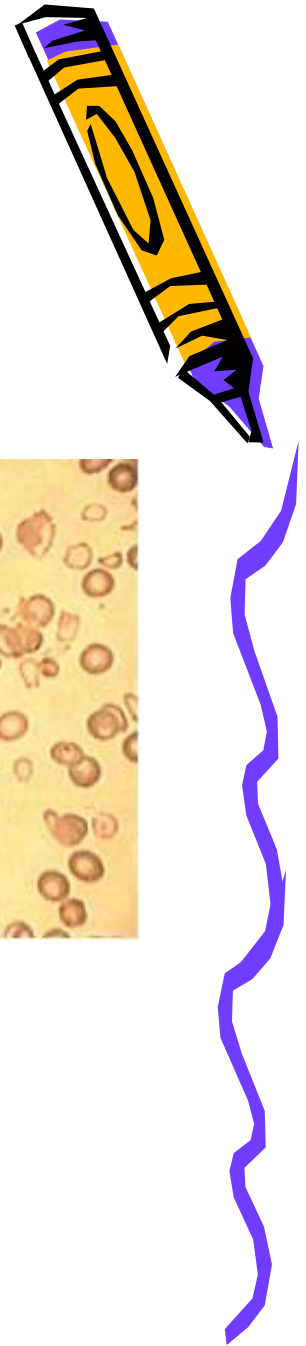
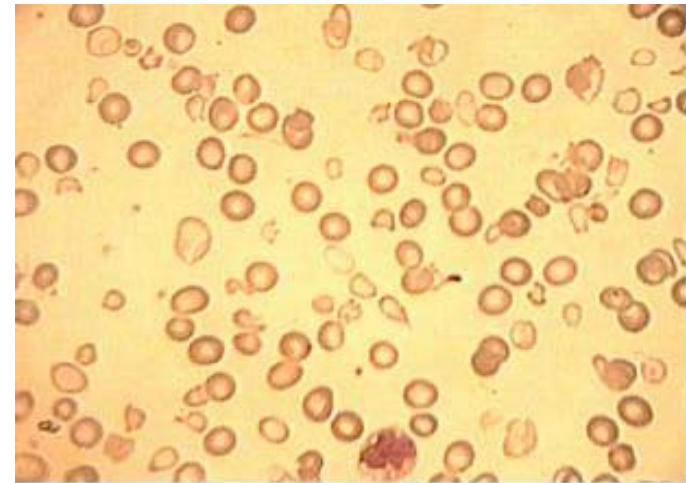


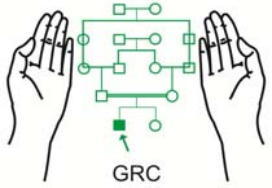
- Age: 2 years
- Sex: Female
- Ethnic: Punjabi
- Consanguinity: 1st cousins
- Complaints:
  - Progressive pallor since 3 months of age
  - Fever off and on 1 month
- Transfusions: Multiple
- Examination:
  - Liver: 2 cm
  - Spleen: 4 cm





- 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





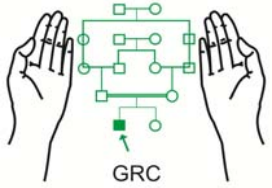
### Father

- 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

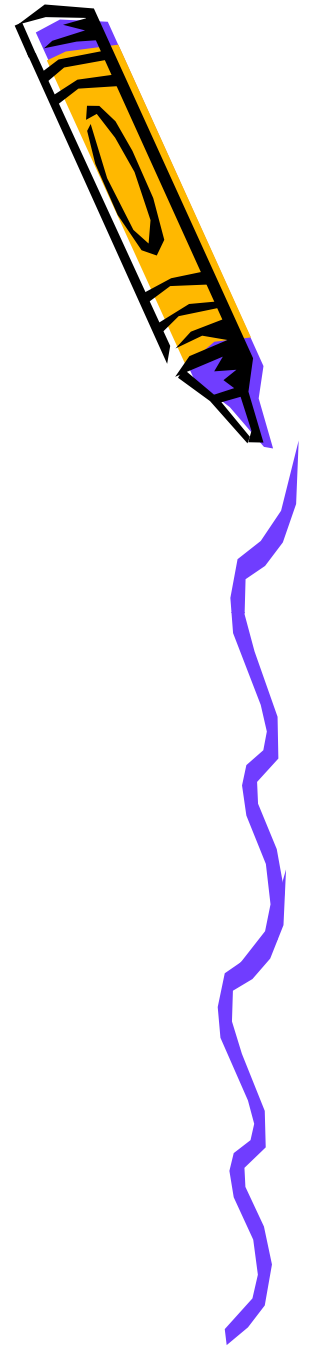
### Mother

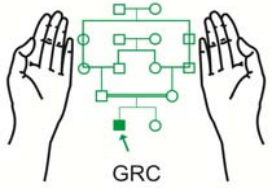
- WBC:  $6.8 \times 10^9/L$
- RBCs:  $5.48 \times 10^{12}/L$
- Hb: 10.6 g/dl
- MCV: 64.5 fl
- MCV: 19.2 pg
- Platelets: 270



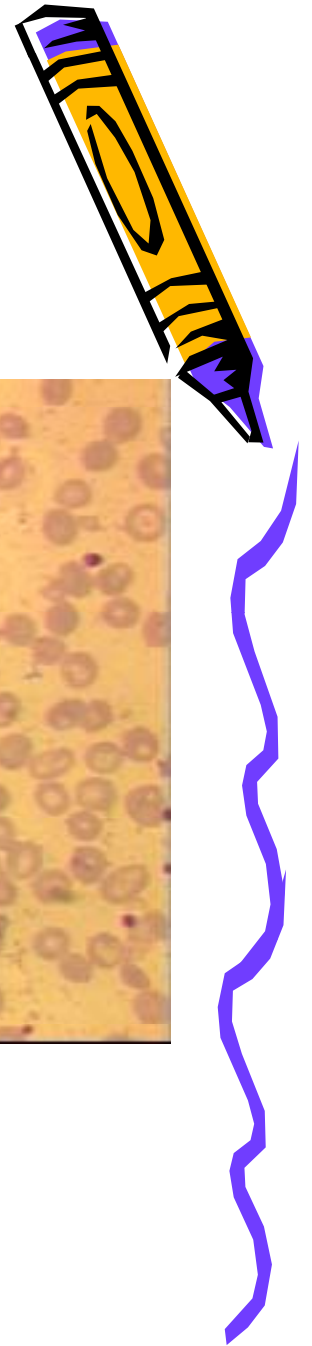
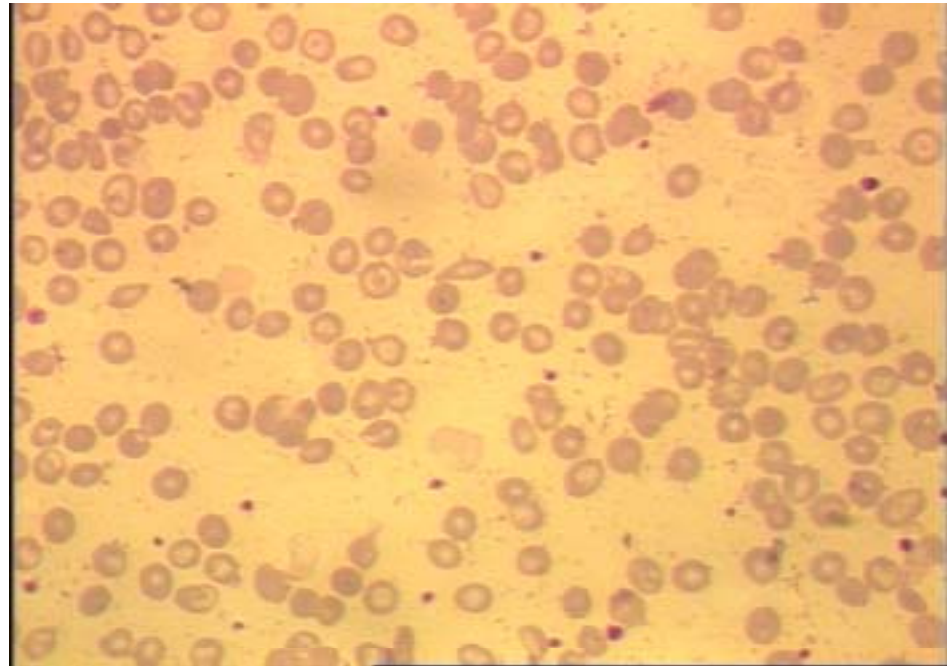


- Age: 16 years
- Sex: Male
- Ethnic: Pathan (Bhitani)
- Consanguinity: 1st cousins
- Complaints:
  - Progressive pallor 6 years
  - Ill health 2 years
- Transfusions: Ten times
- Examination:
  - Liver: 2 cm
  - Spleen: 8 cm

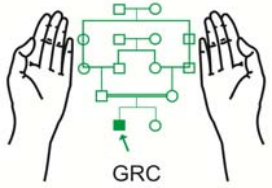




- WBC:  $7.23 \times 10^9/L$
- RBCs:  $3.37 \times 10^{12}/L$
- Hb: 7.4 g/dl
- MCV: 70.0 fl
- MCV: 22.2 pg
- Platelets: 198
- Reticulocytes: 3.5%
- Hb-F: 15.3%
- Hb-A2: 4.6%





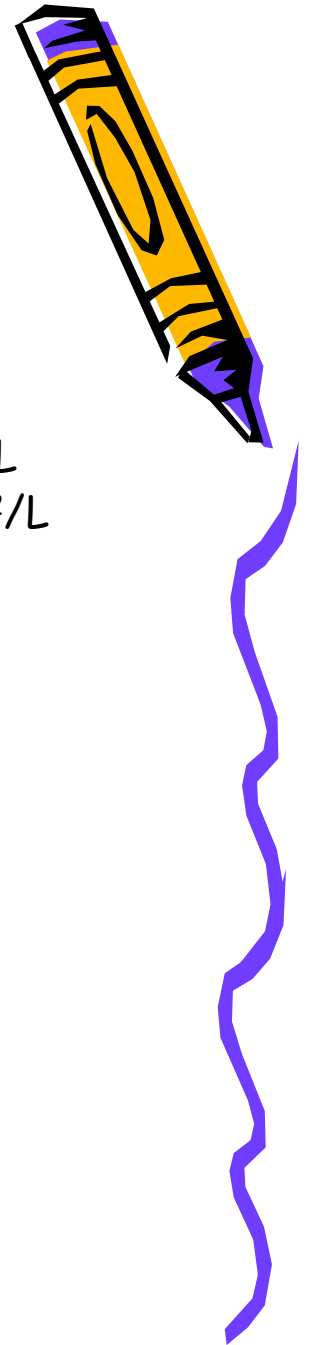
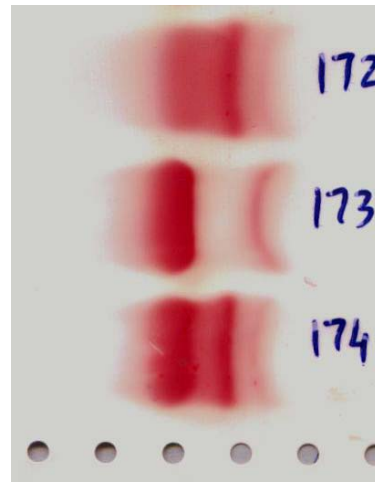


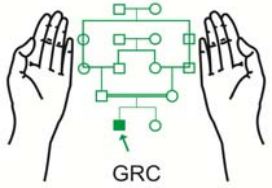
### Father (173)

- 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

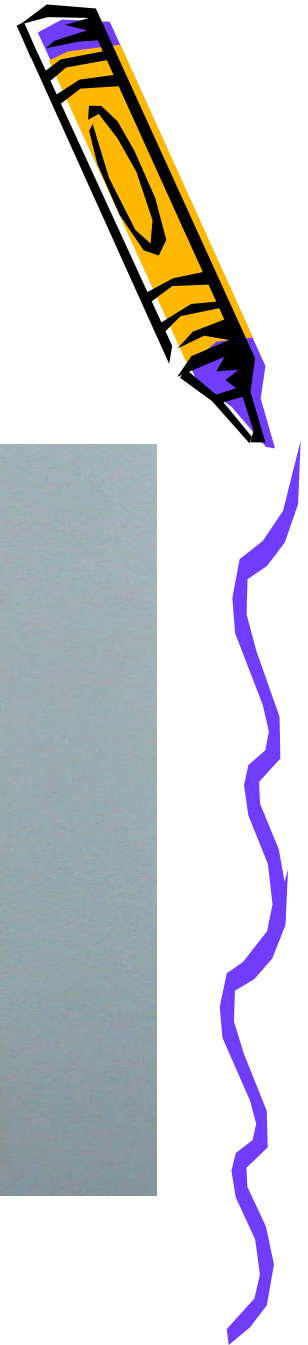
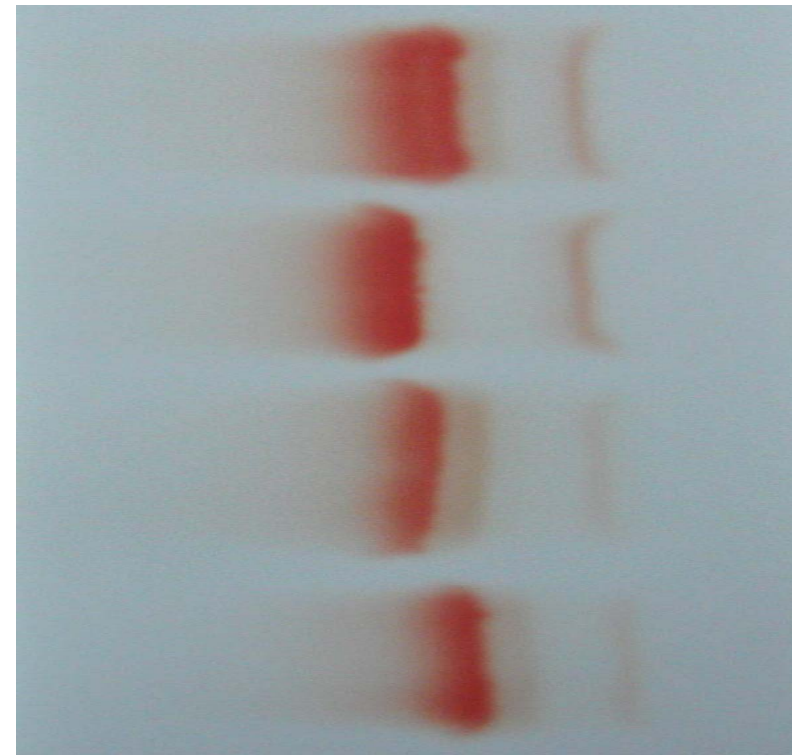
### Mother (174)

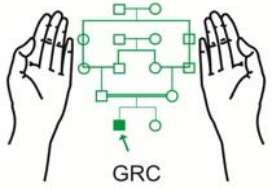
- WBC:  $6.8 \times 10^9/L$
- RBCs:  $4.18 \times 10^{12}/L$
- Hb: 10.6 g/dl
- MCV: 84.5 fl
- MCV: 27.2 pg
- Platelets: 270



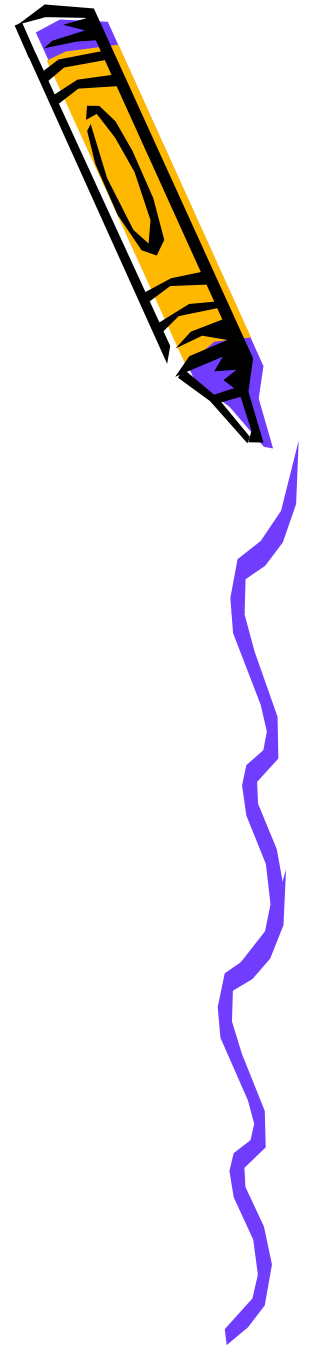


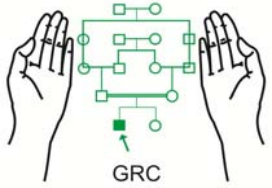
- Age: 1 year
- Sex: Male
- Ethnic: Pathan
- Consanguinity: 1st cousins
- Complaints:
  - Cyanosis since birth
- WBC:  $7.23 \times 10^9/L$
- RBCs:  $4.23 \times 10^{12}/L$
- Hb: 10.4 g/dl
- MCV: 79.0 fl
- MCV: 26.2 pg
- Platelets: 298
- Reticulocytes: 4.5%



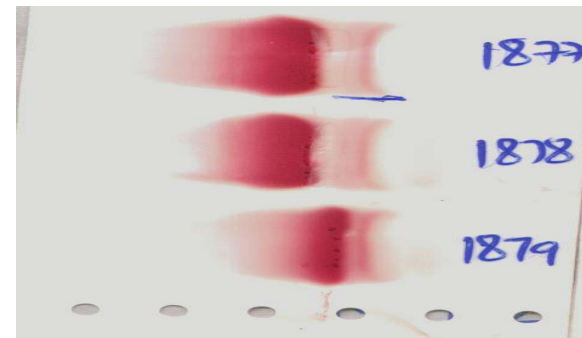
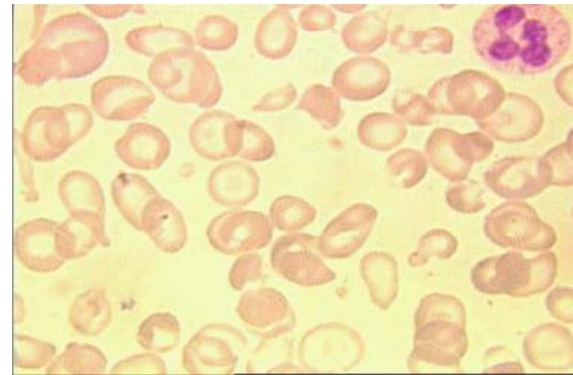


- Age: 17 years
- Sex: Female
- Ethnic: Punjabi
- Consanguinity: Unrelated
- Complaints: None
- Transfusions: Nil
- Examination:
  - Liver: NP
  - Spleen: NP



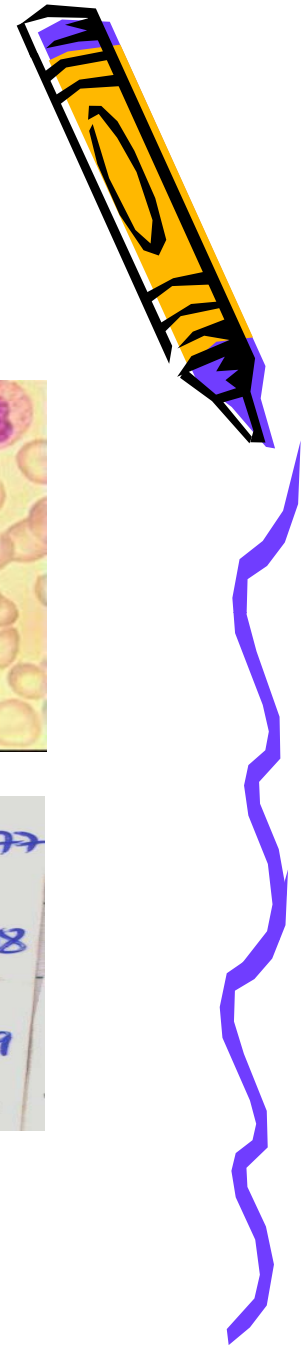


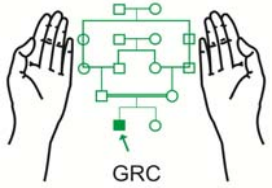
- WBC:  $7.73 \times 10^9/L$
- RBCs:  $5.21 \times 10^{12}/L$
- Hb: 9.4 g/dl
- MCV: 56.5 fl
- MCV: 18.2 pg
- Platelets: 241
- Reticulocytes: 2.6%



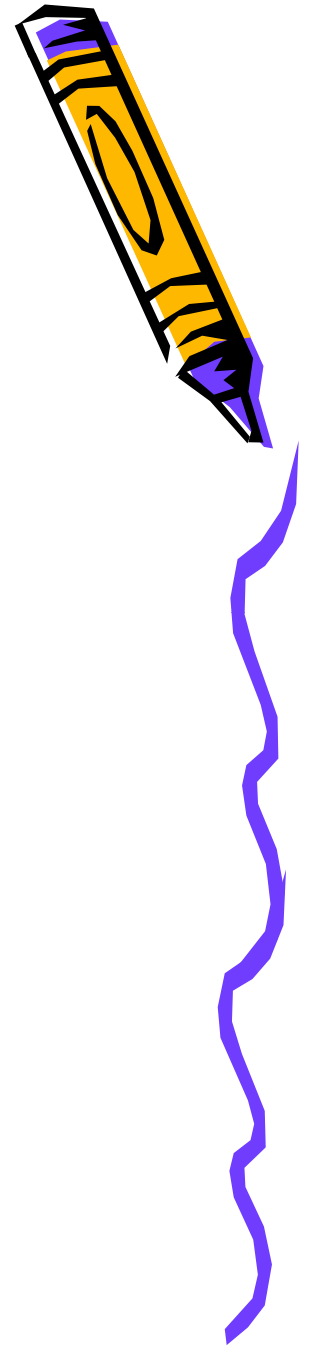
Hb-A2: 4.5%

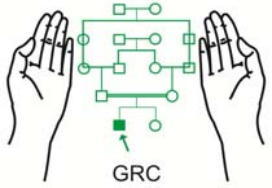
PCR: Hb-D/Fr 8-9



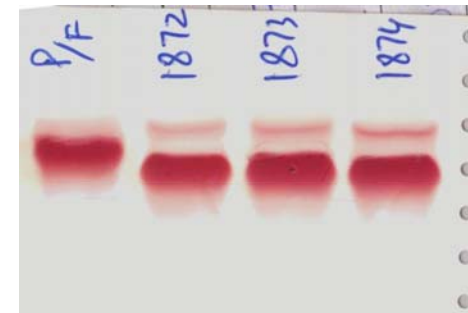
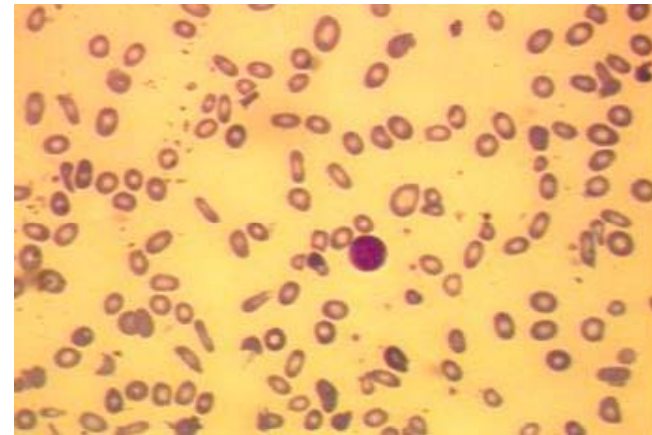


- Age: 47 years
- Sex: Female
- Ethnic: Punjabi
- Consanguinity: 1<sup>st</sup> cousins
- Complaints:
  - Weakness: 2 months
- Transfusions: None
- Examination:
  - Liver: NP
  - Spleen: NP



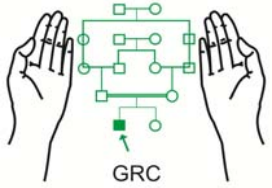


- WBC: 4.75 X 10<sup>9</sup>/L
- RBCs: 2.29 X 10<sup>12</sup>/L
- Hb: 5.2 g/dl
- MCV: 73.8 fl
- MCV: 22.7 pg
- Platelets: 157
- Reticulocytes: 3.0%

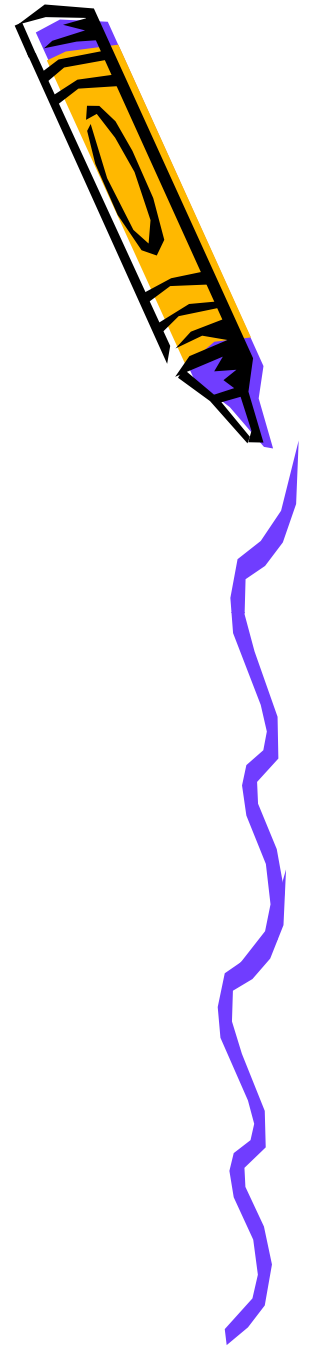


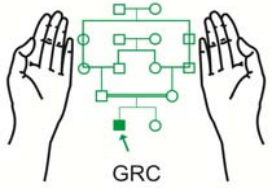
Hb-A2: 5.3%  
PCR: Het Cd5



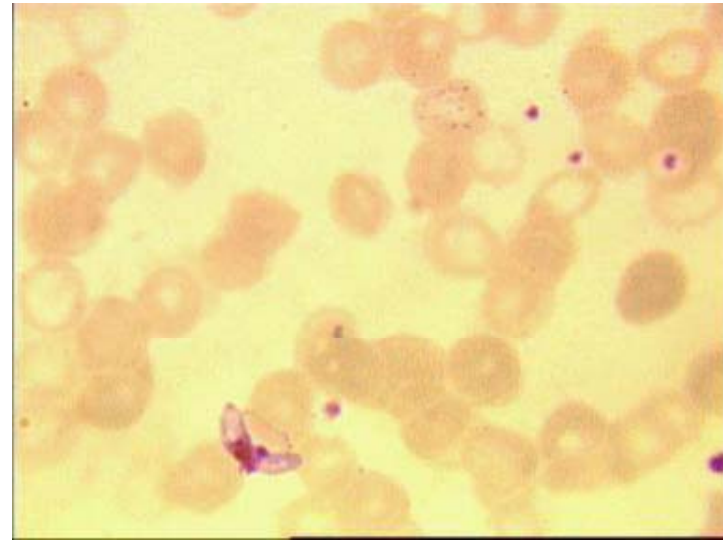


- Age: 26 years
- Sex: Male
- Ethnic: Pathan
- Consanguinity: 1st cousins
- Complaints:
  - Fever with chills 1 week
- Transfusions: Nil
- Examination:
  - Liver: NP
  - Spleen: 2 cm

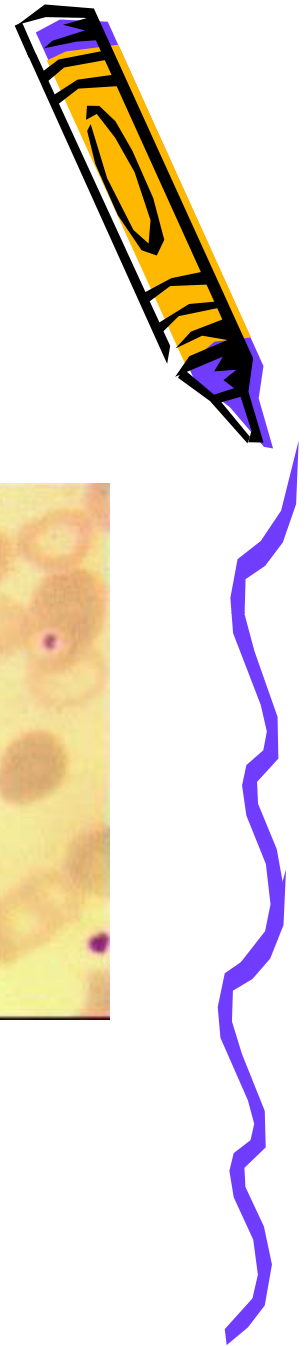




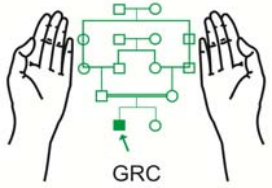
- WBC:  $13.23 \times 10^9/L$
- RBCs:  $4.21 \times 10^{12}/L$
- Hb: 10.5 g/dl
- MCV: 61.5 fl
- MCV: 19.3 pg
- Platelets: 171
- Reticulocytes: 4.2%



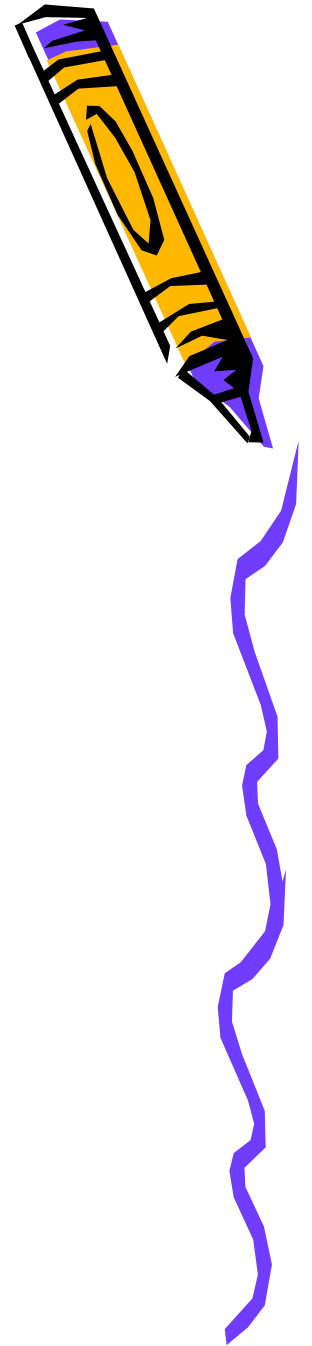
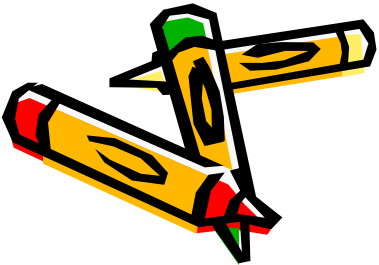
Hb-A2: 5.1%  
PCR: Fr 8-9

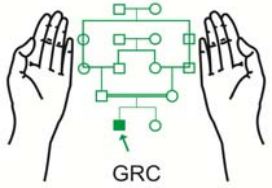




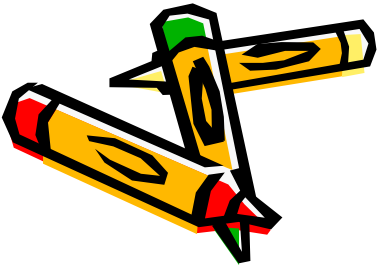
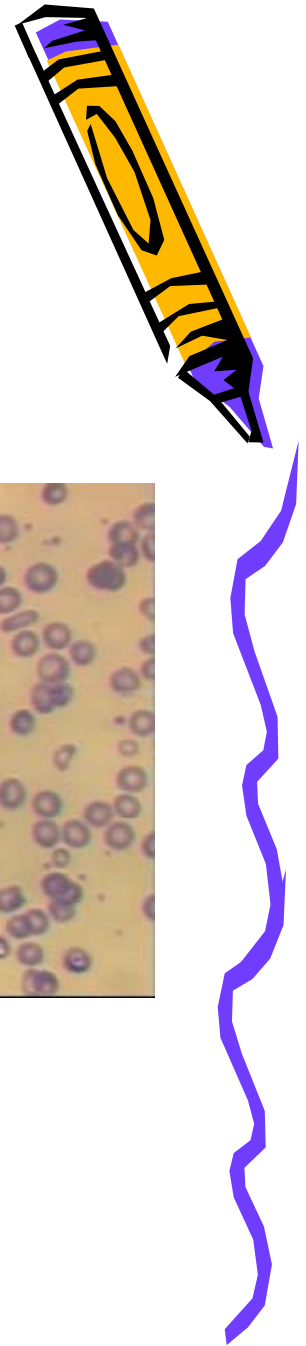
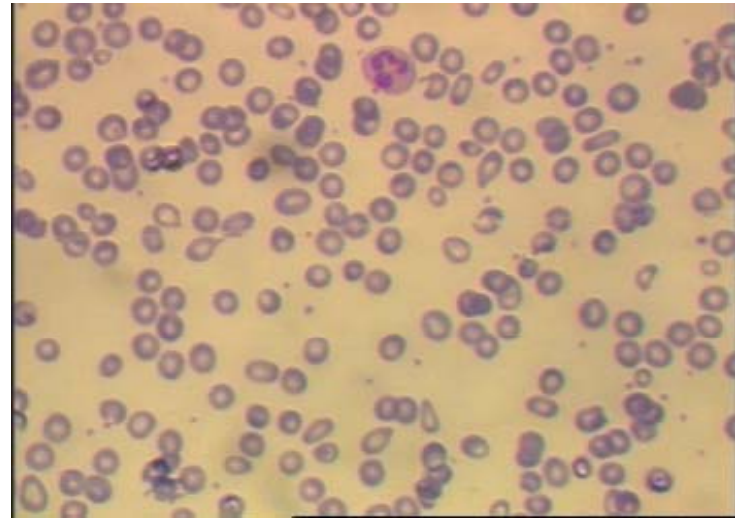


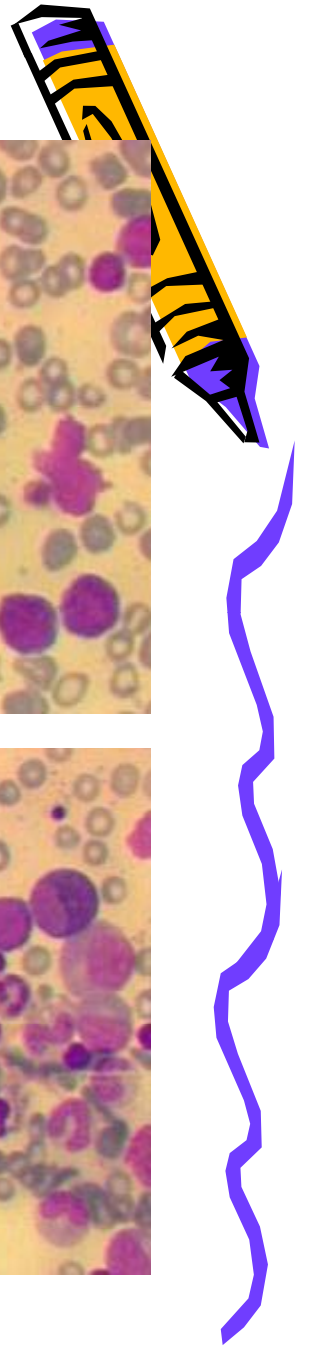
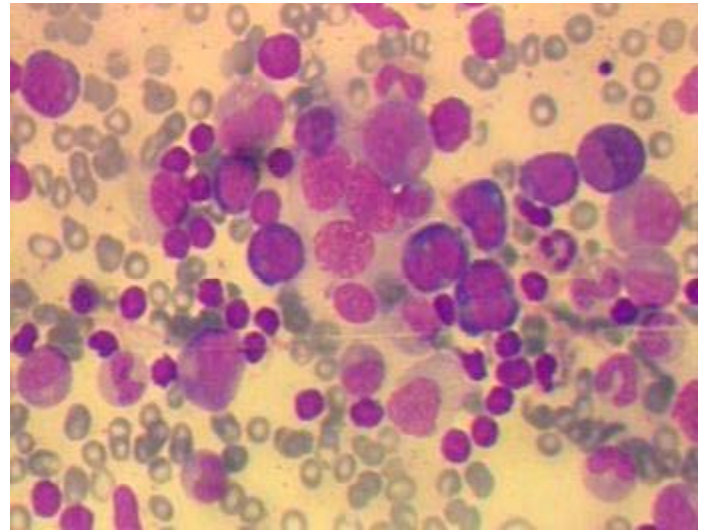
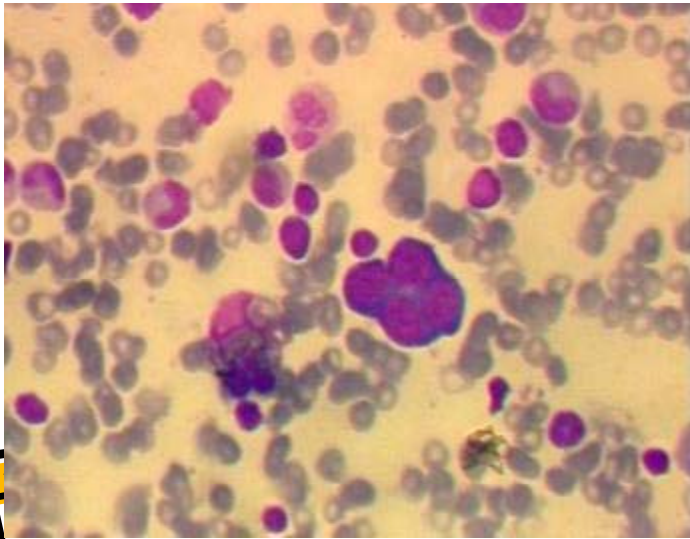
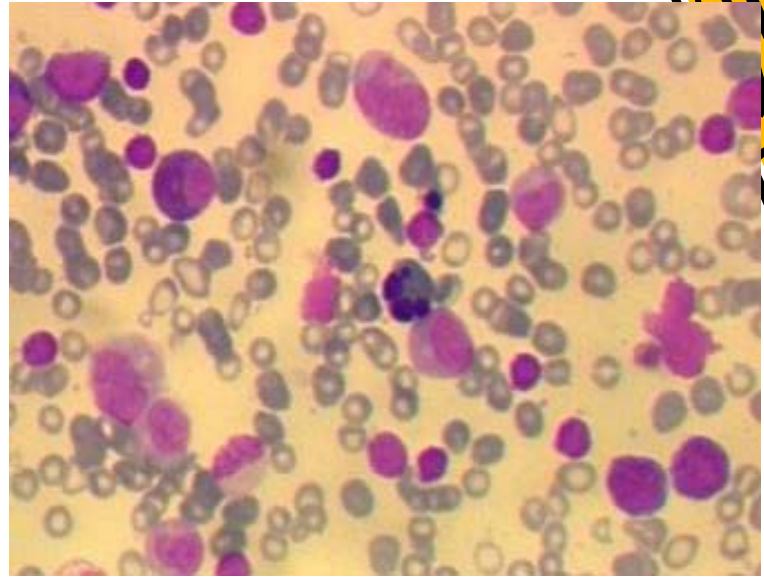
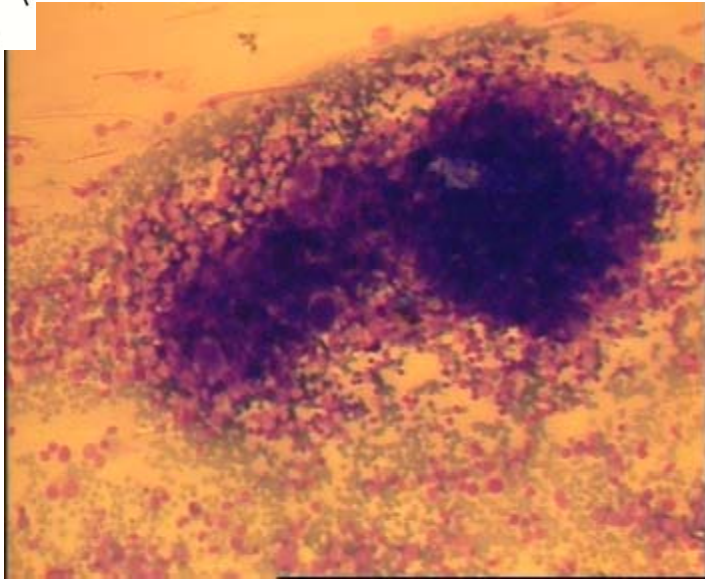
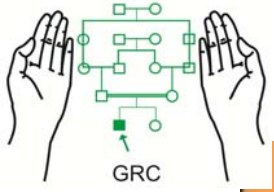
- Age: 1 year
- Sex: Male
- Ethnic: Punjabi
- Consanguinity: 1<sup>st</sup> cousins
- Complaints:
  - Progressive pallor 6 months
  - Fever off and on 6 months
- Transfusions: Thrice
- Examination:
  - Liver: Nil
  - Spleen: tip

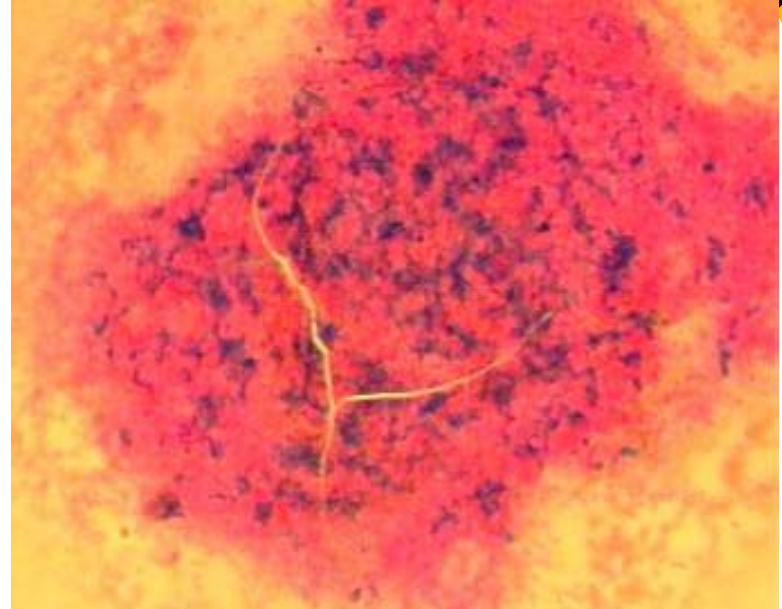
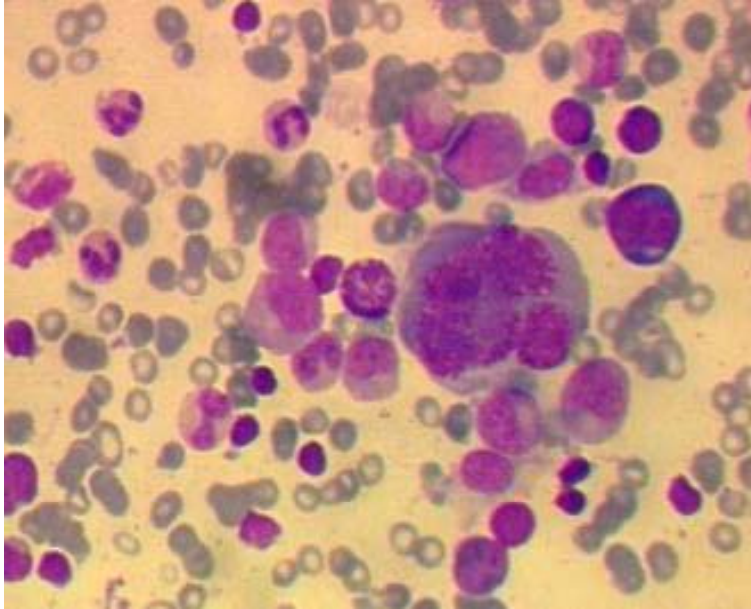
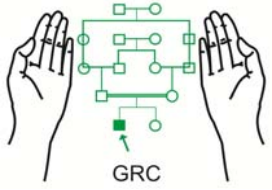




- WBC:  $13.23 \times 10^9/L$
- RBCs:  $3.21 \times 10^{12}/L$
- Hb: 8.4 g/dl
- MCV: 78.5 fl
- MCH: 26.2 pg
- Platelets: 341
- Reticulocytes: 0.2%
- Hb-F: 0.9%
- Hb-A2: 2.9%
- Electrophoresis: No abnormal band seen

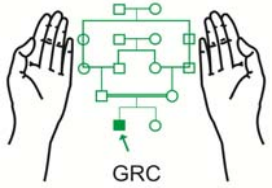




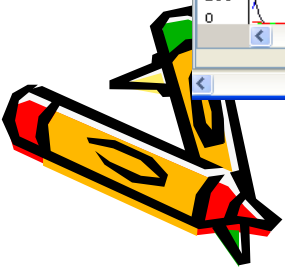
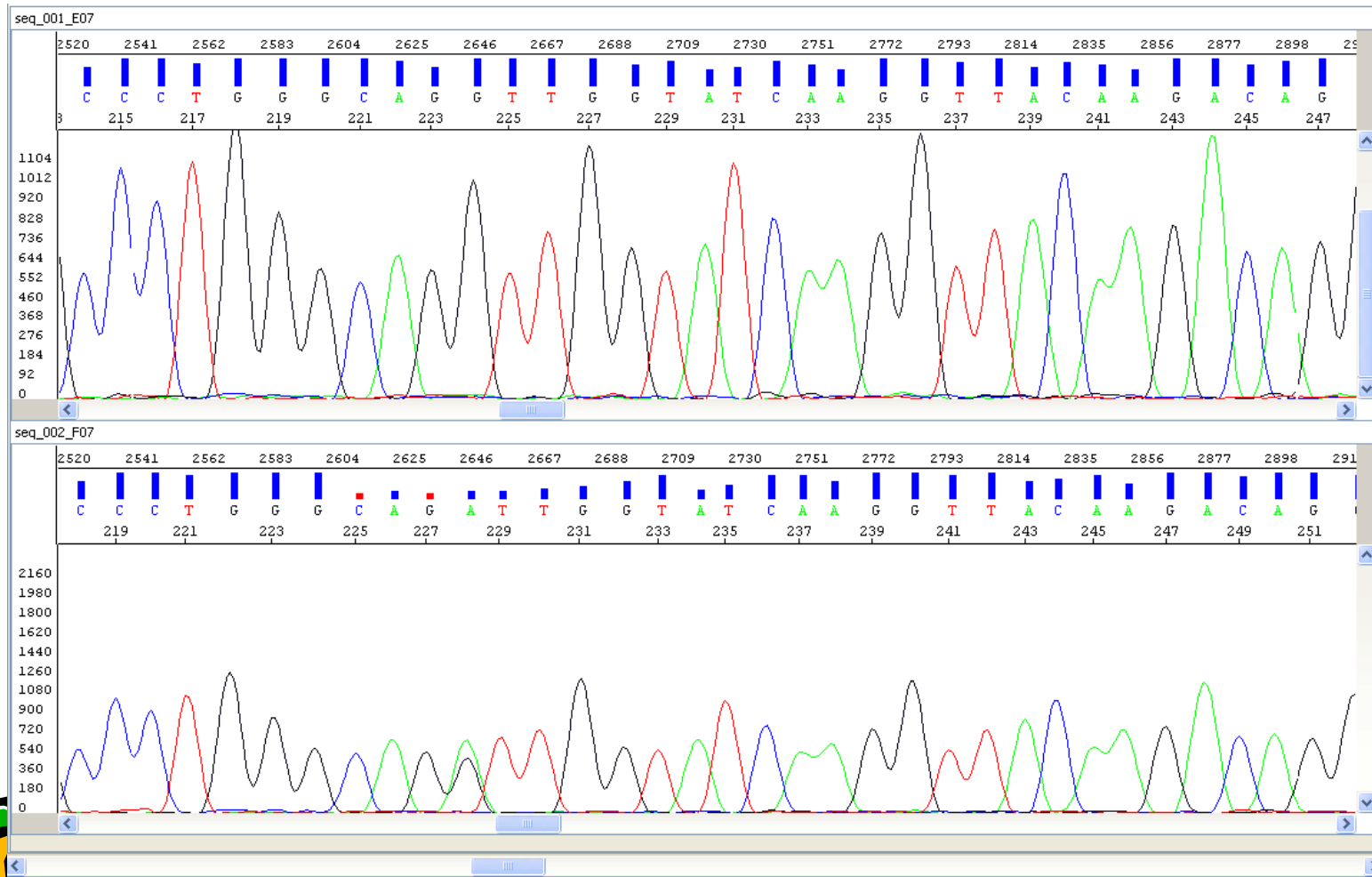
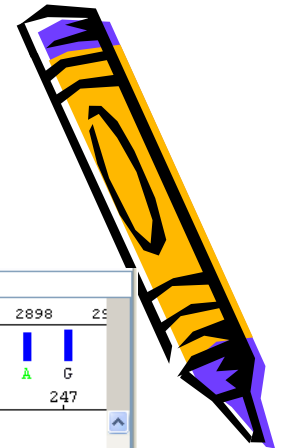


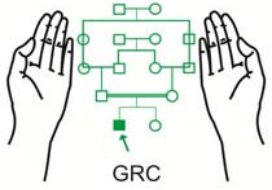
PCR: Heterozygous del 619



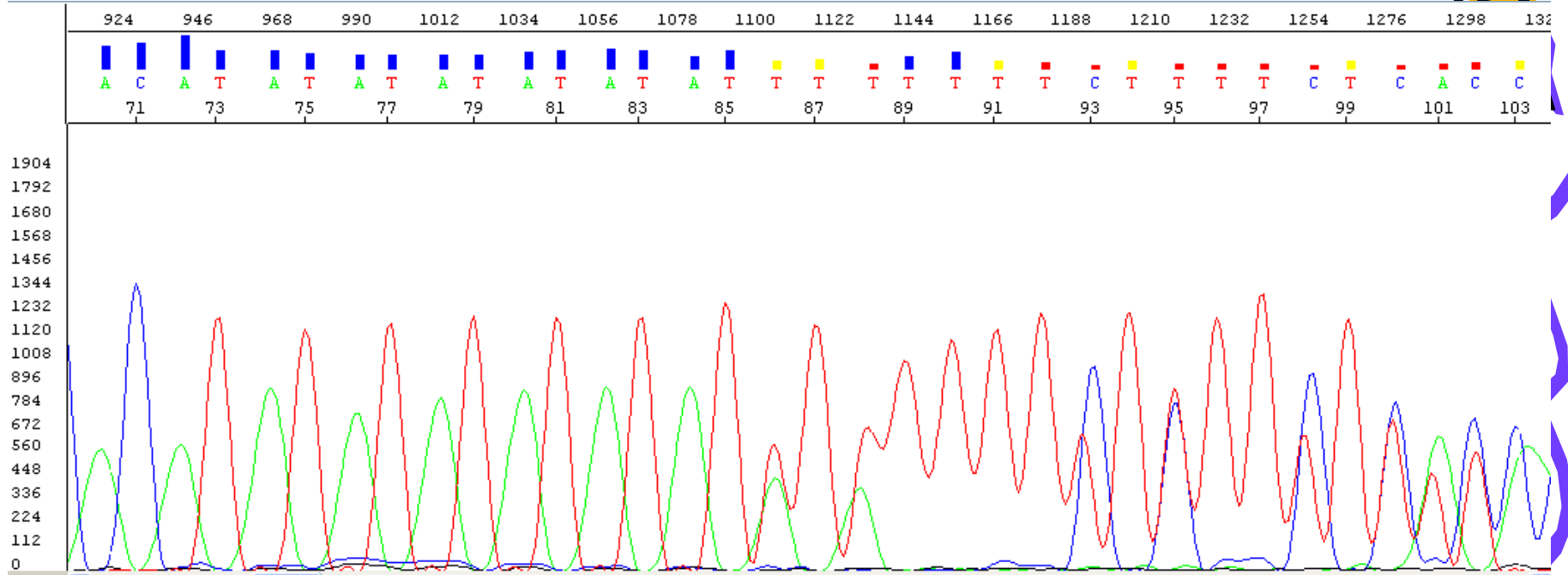
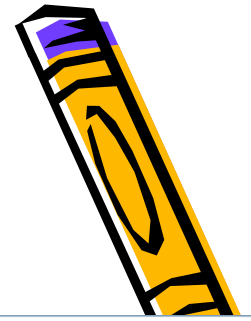


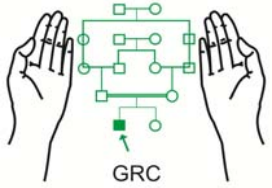
# IVSI-1 G-A





**-527 (+ATA)**





# Cd 45 (+T)

