Pediatric Congenital Hematologic Disorders Research Center
مرکزتحقیقات بیماری های خونی مادرزادی کودکان

Case Studies
Hemoglobinopathies

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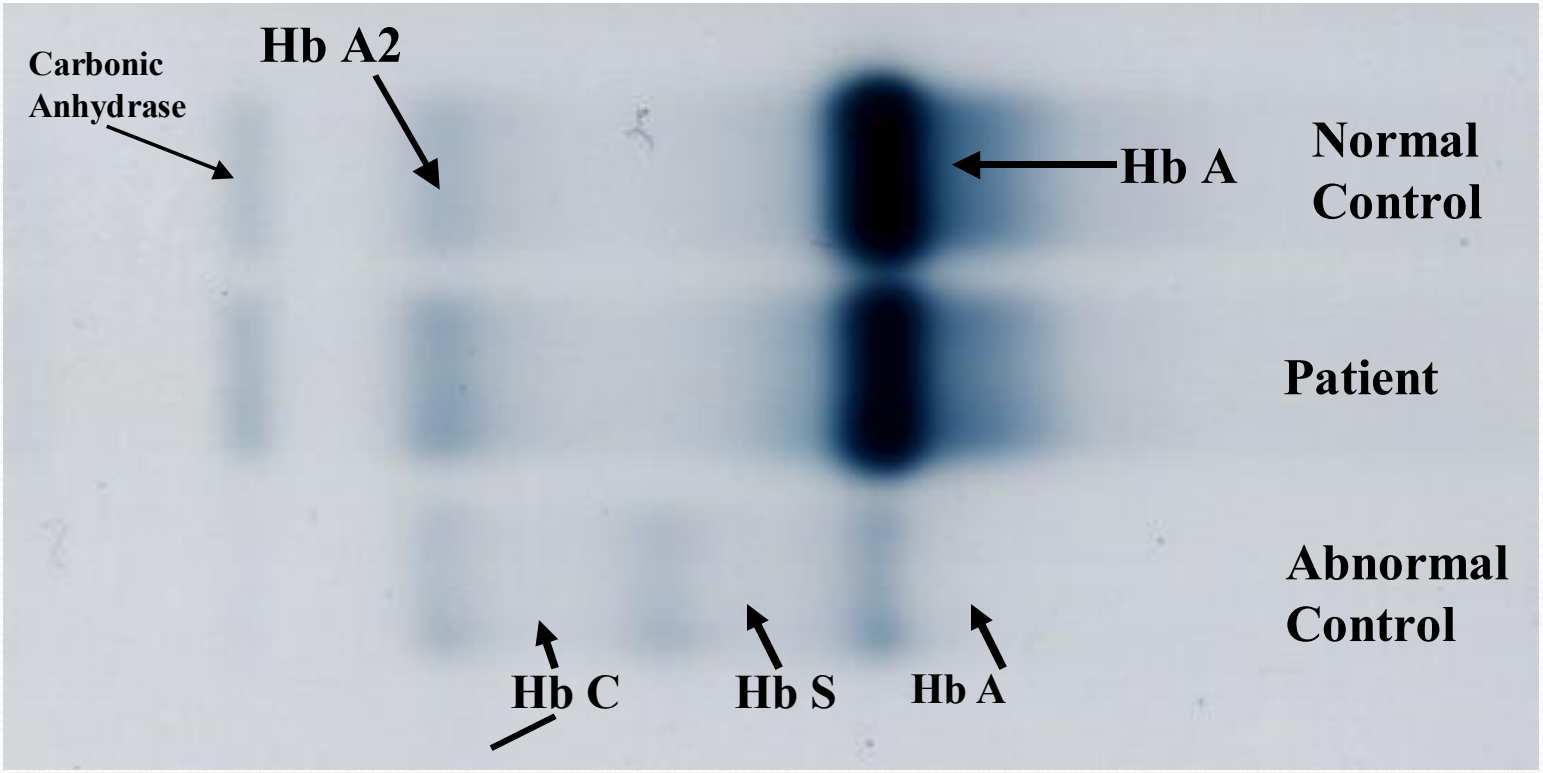
Haemoglobin Electrophoresis at alkaline pH



Marks position of HbA2

(Using abnormal Hbs as markers)

Case 1

18 year old young man seen for a medical examination prior to immigration

- **Past medical history unremarkable.**
- **Family of Sicilian descent.**
- **Physical examination is normal**

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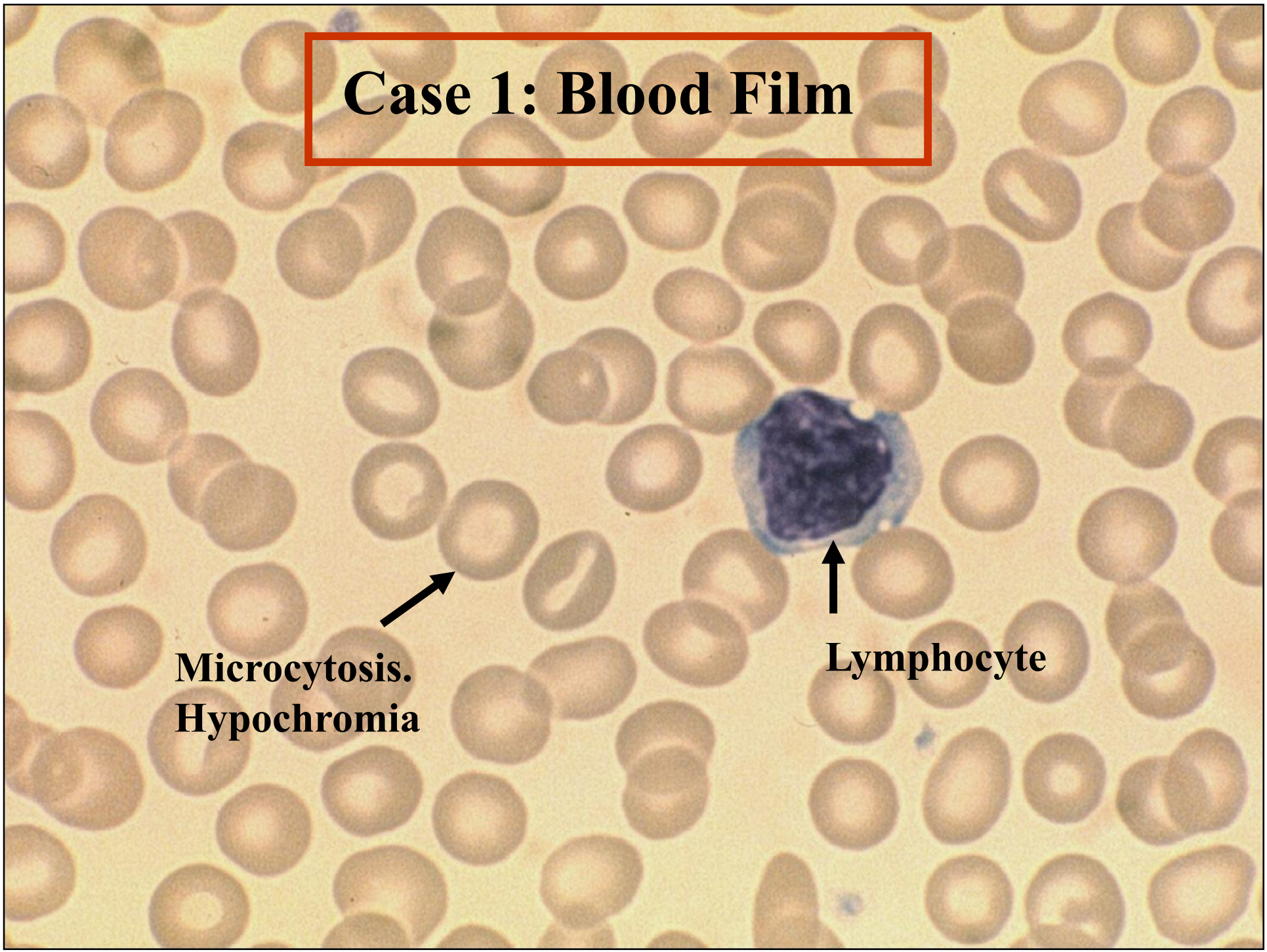
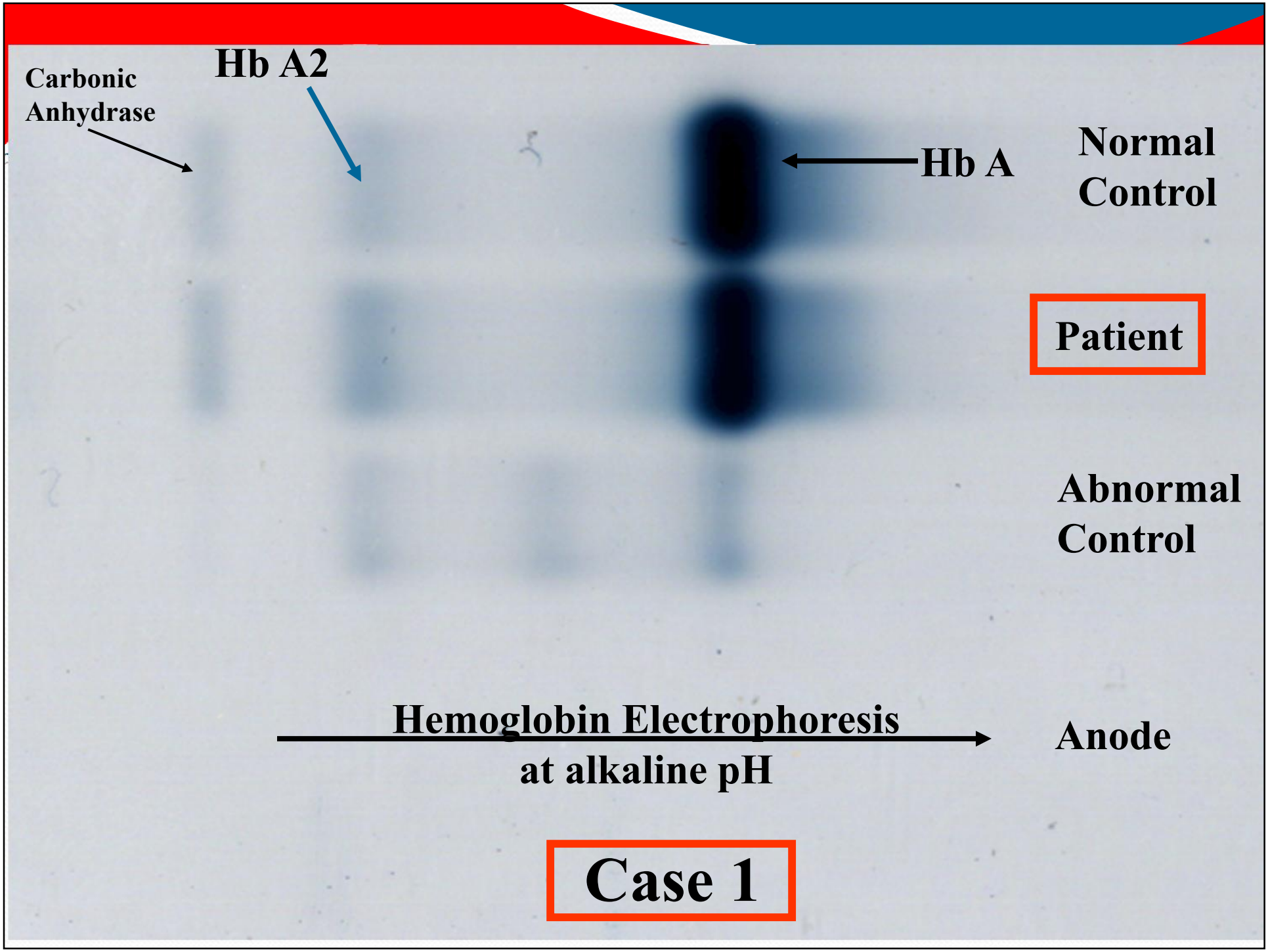
Case 1

- Hb 132 g/l (140-180)
- MCV 66.1 fl (80-100)

Case 1: Blood Film




Carbonic Anhydrase
Hb A2
Hb A
Normal Control
Patient
Abnormal Control
Hemoglobin Electrophoresis at alkaline pH
Anode
Case 1

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Case 1

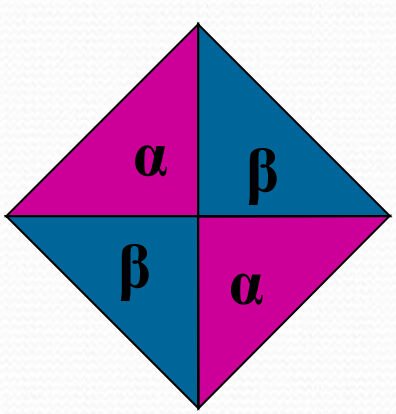
Haemoglobin Electrophoresis:

Hb A	**94.0%**
Hb F	**<1.5%**
Hb A_2	**5.2%**

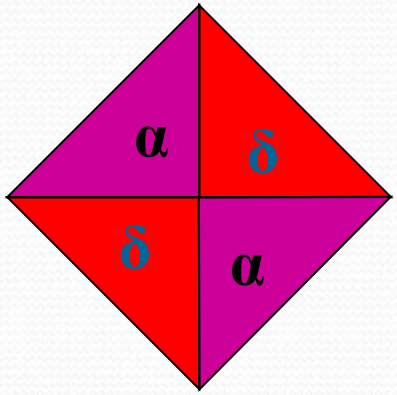
Diagnosis: β Thalassemia trait

Genotype ααβ/ααβ or ααβ/αα-

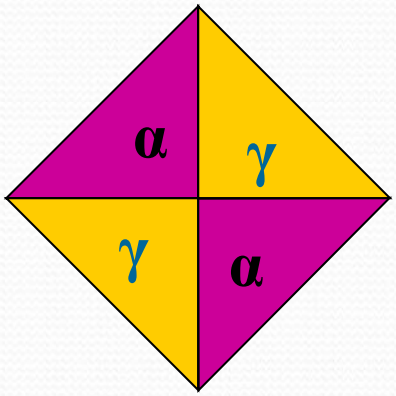
Haemoglobins Produced



HbA



Hb A2



Hb F

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Case 2

- 6 year old African American girl
- Admitted to hospital with abdominal pain and fever.
- Past history : swelling of hands and feet at age 1
  :previous episodes of abdominal pain
- Physical examination:
  - pallor
  - mild jaundice
  - hepatomegaly
  - no evidence of infection
- Parents healthy

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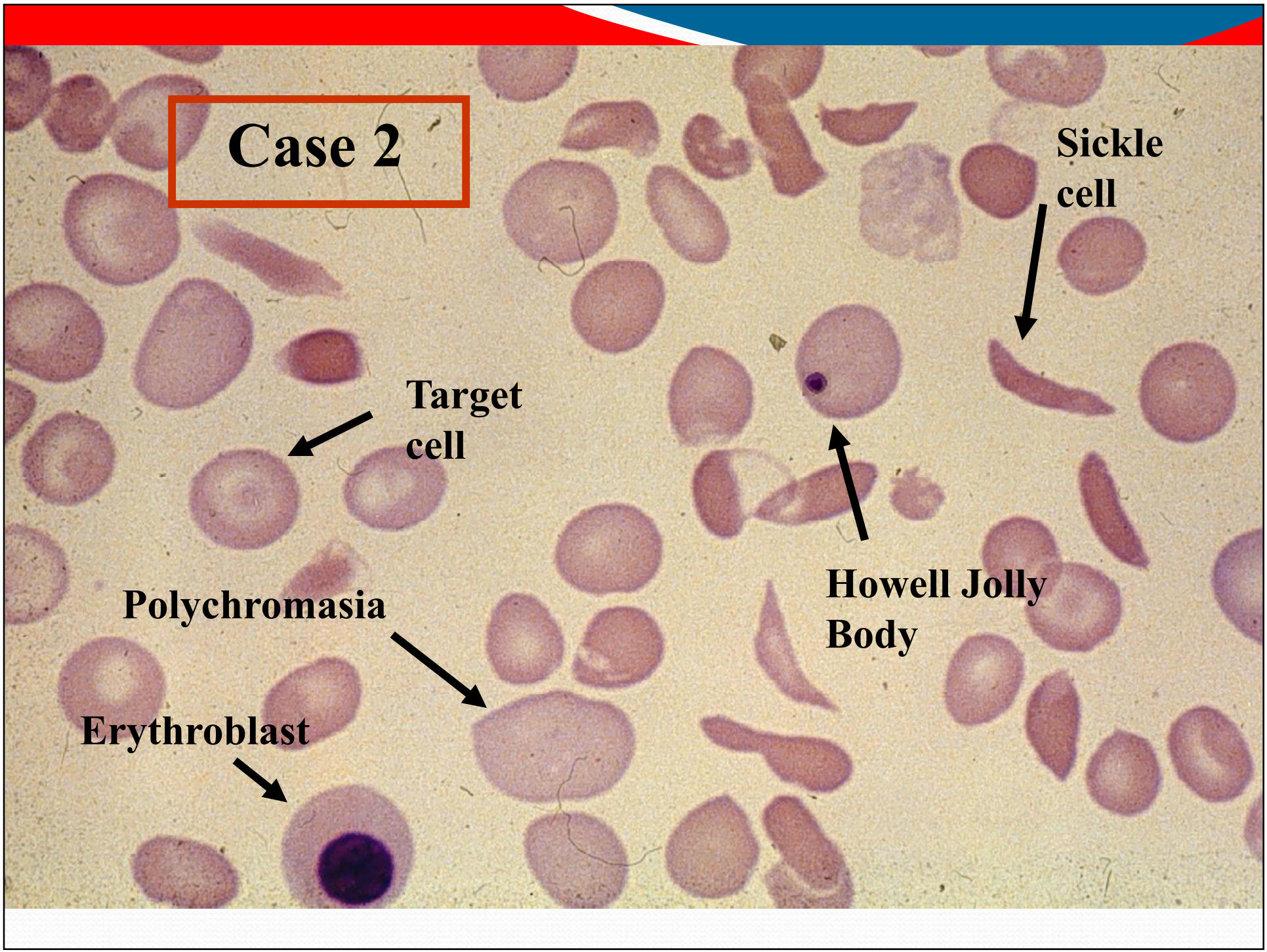
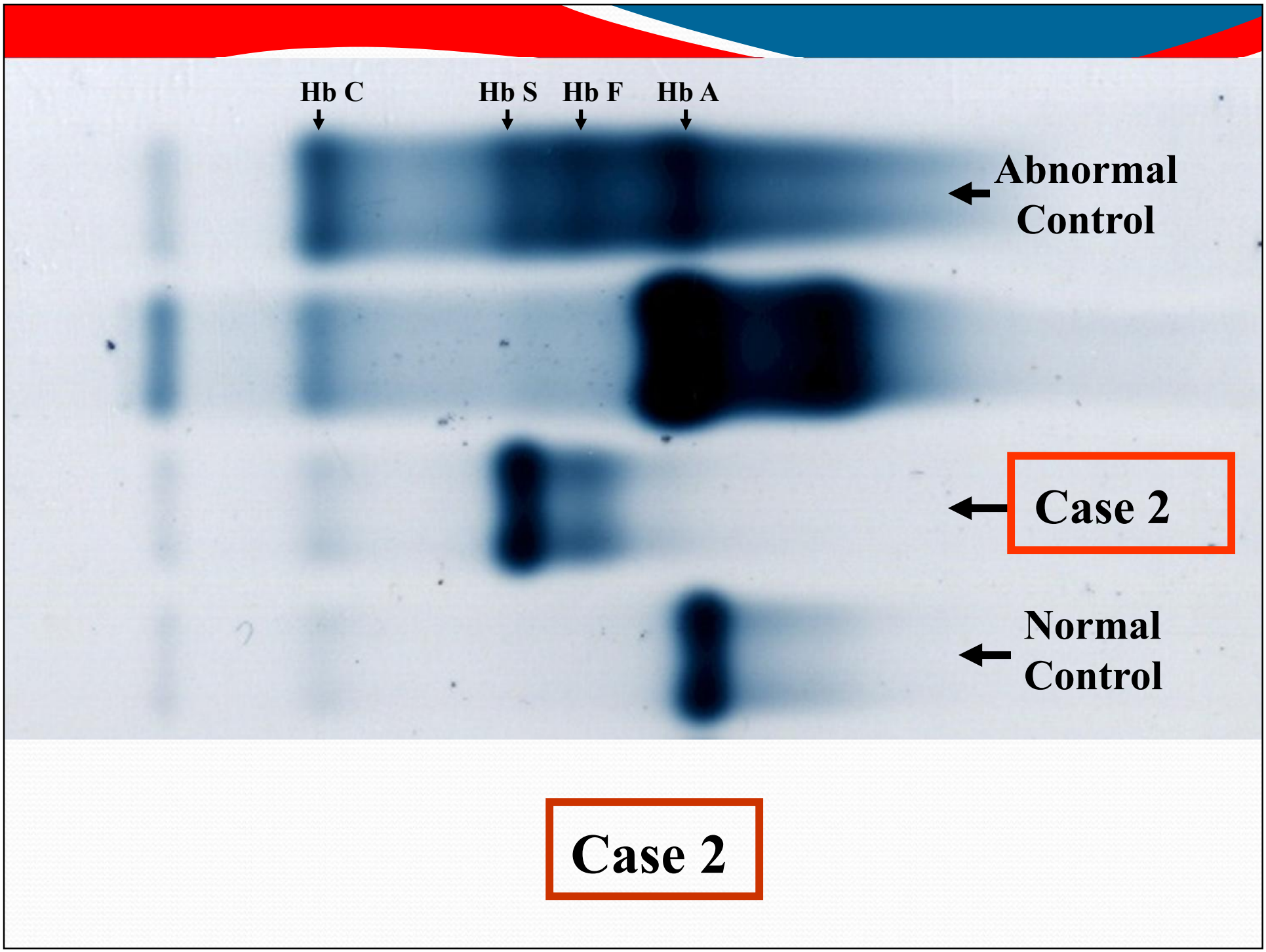
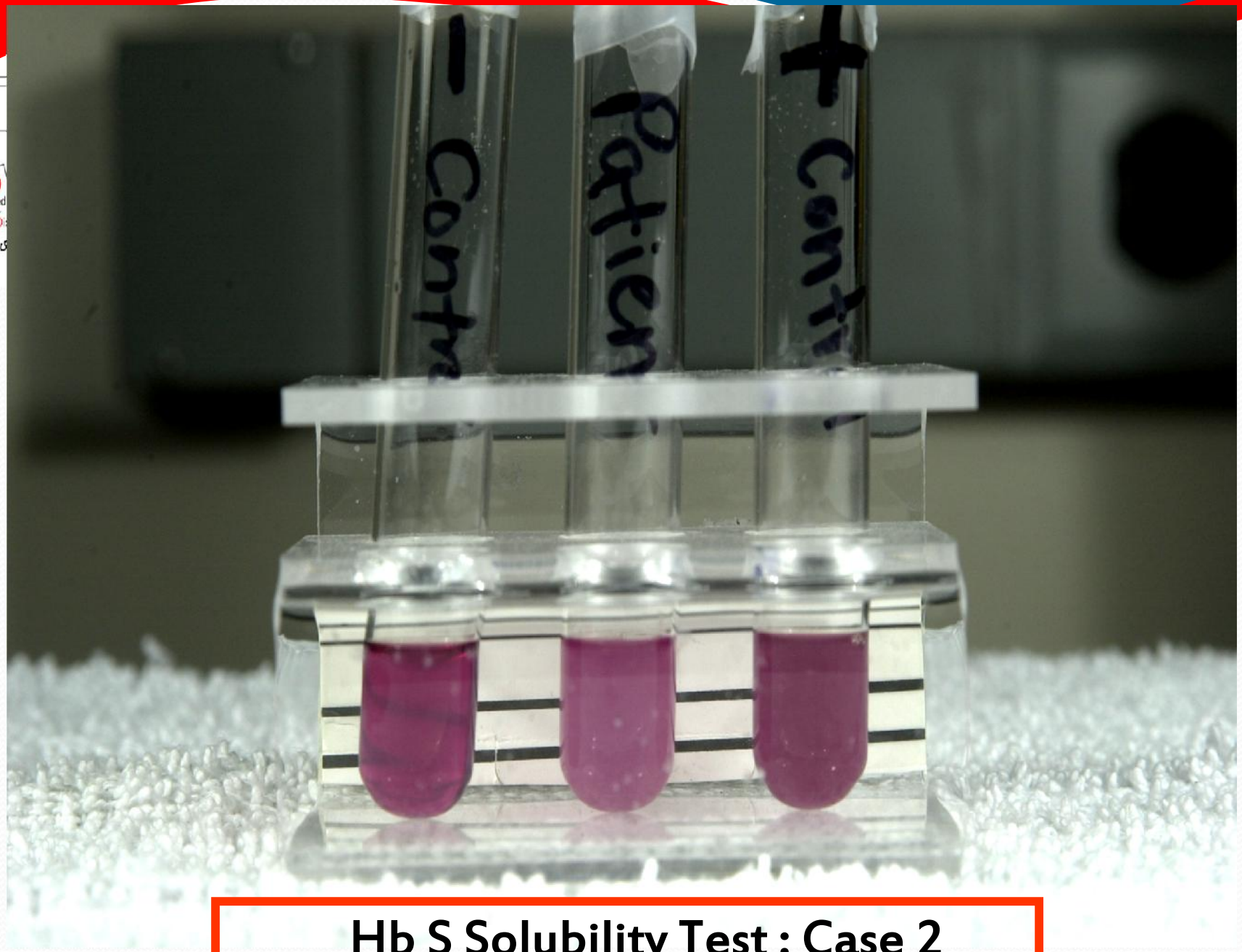
Case 2

Hb	**84 g/L (115-155)**
MCV	**86.5 fl (77-95)**


Case 2
Sickle cell
Target cell
Howell Jolly Body
Polychromasia
Erythroblast



Case 2



Hb S Solubility Test : Case 2

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Case 2

Haemoglobin Electrophoresis:

Hb A 0%
Hb S 87%
Hb F 9.7%
Hb A_2 3.3%

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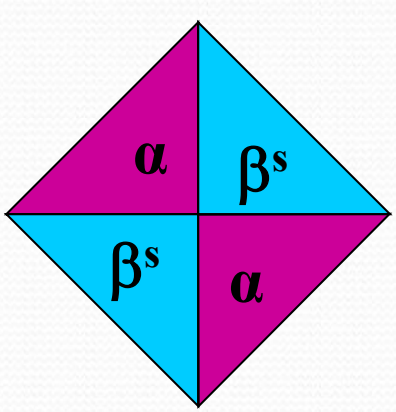
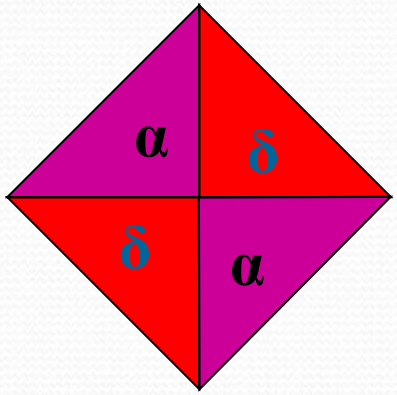
Diagnosis : Hb SS Disease

Genotype $\alpha\alpha\beta^s/\alpha\alpha\beta^s$

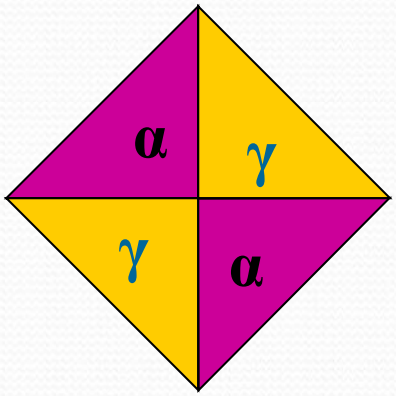
Haemoglobins Produced :



HbSS



Hb A2



Hb F

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Case 3

6 month old baby girl of Italian ancestry

- **Failure to thrive and pallor**
- **Family History:**
  - **no definite history of a blood problem**
  - **mother "anaemic" during pregnancy; given iron**
  - **one sibling :history of mild anaemia; given iron**
- **Physical Examination:**
  - **pallor**
  - **hepatosplenomegaly**

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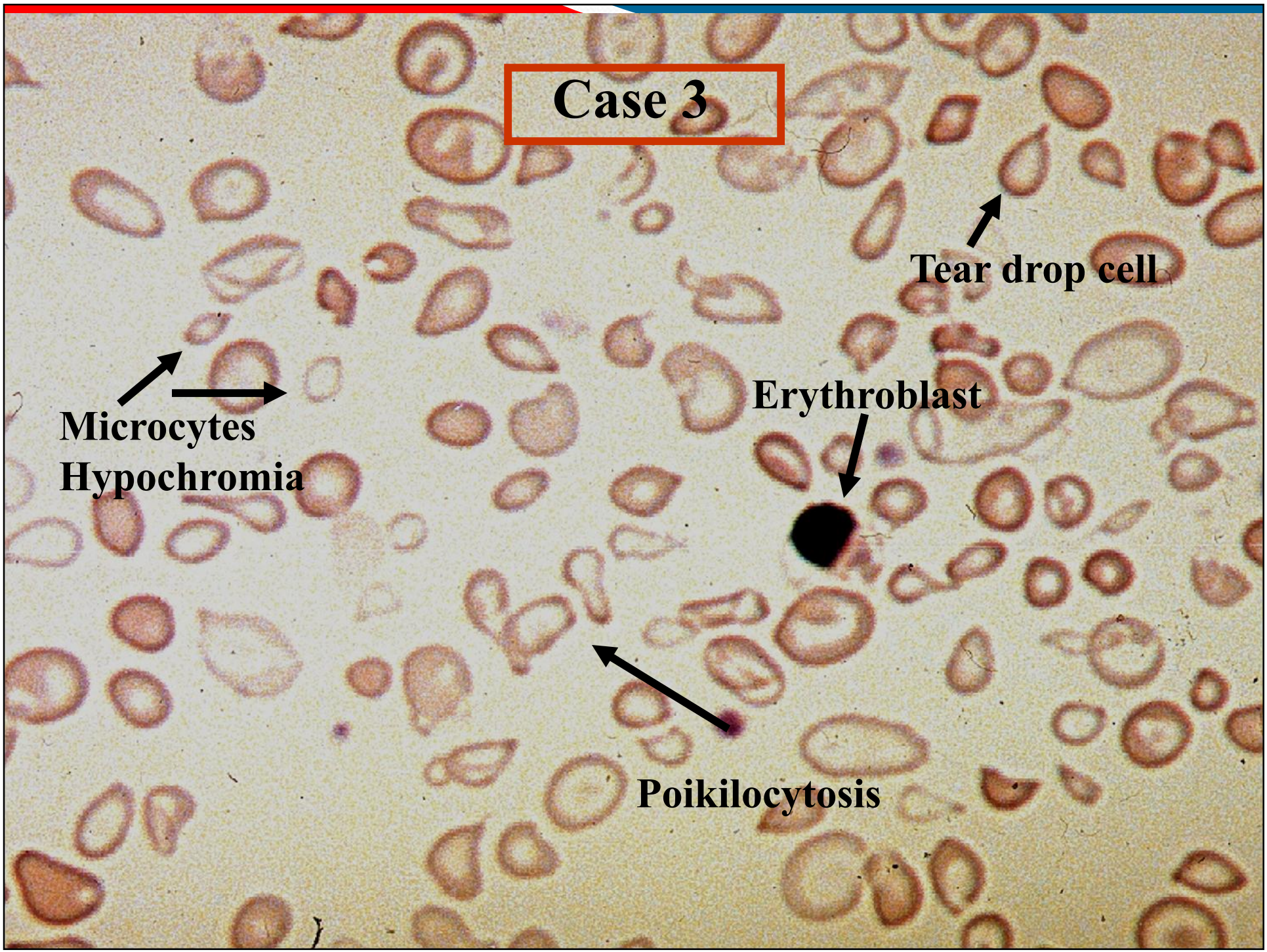
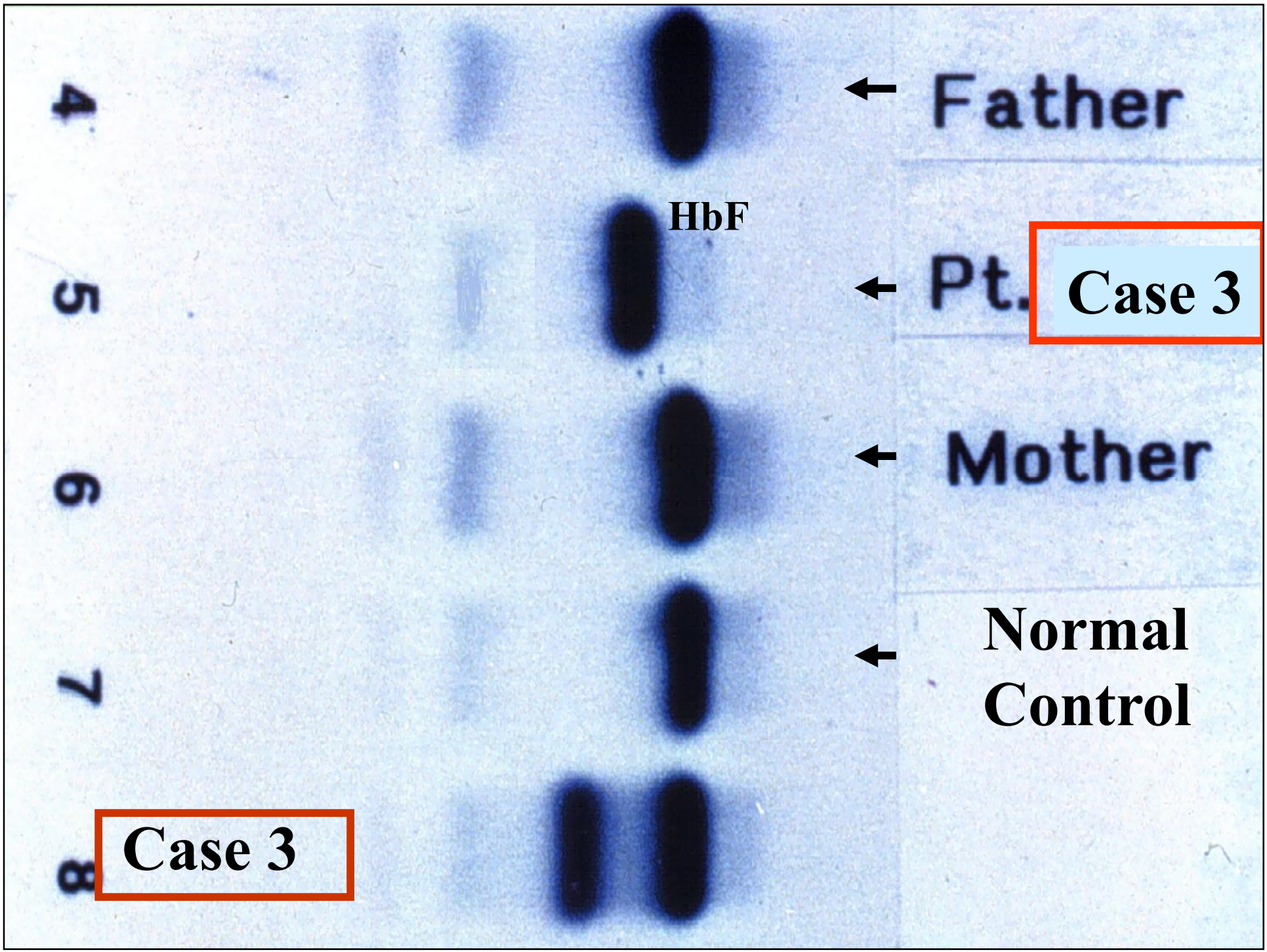
Case 3

Hb **69 g/L (105-135)**

MCV **68.5 fl (70-86)**


Case 3
Tear drop cell
Erythroblast
Microcytes
Hypochromia
Poikilocytosis


4
5
6
7
8
Case 3
HbF
Father
Pt.
Case 3
Mother
Normal
Control

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Case 3

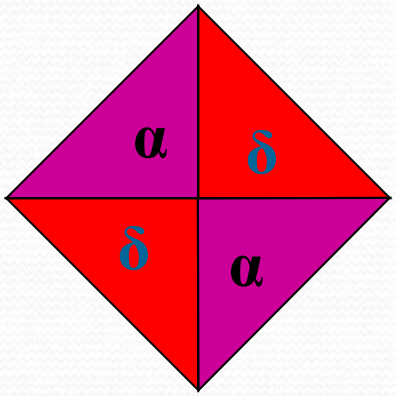
Haemoglobin Electrophoresis:

Hb A	**0%**
Hb F	**98.5%**
Hb A_2	**1.5%**

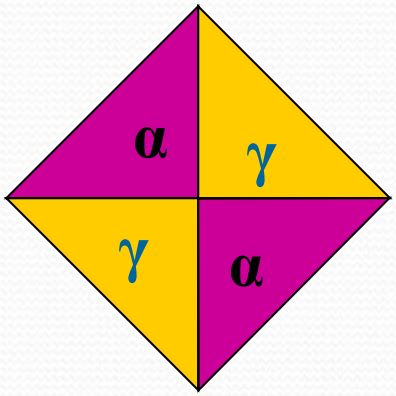
Diagnosis: β Thalassemia major

Genotype αα-/αα-

Haemoglobins Produced



Hb A2



Hb F

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Case 4

- 28 year old Thai woman
- Life long history anaemia and mild jaundice.
- Past history : splenectomy.
- Family History :
  - mother :lifelong microcytic anemia not responsive to iron
  - father and sister: no known history of blood problem.

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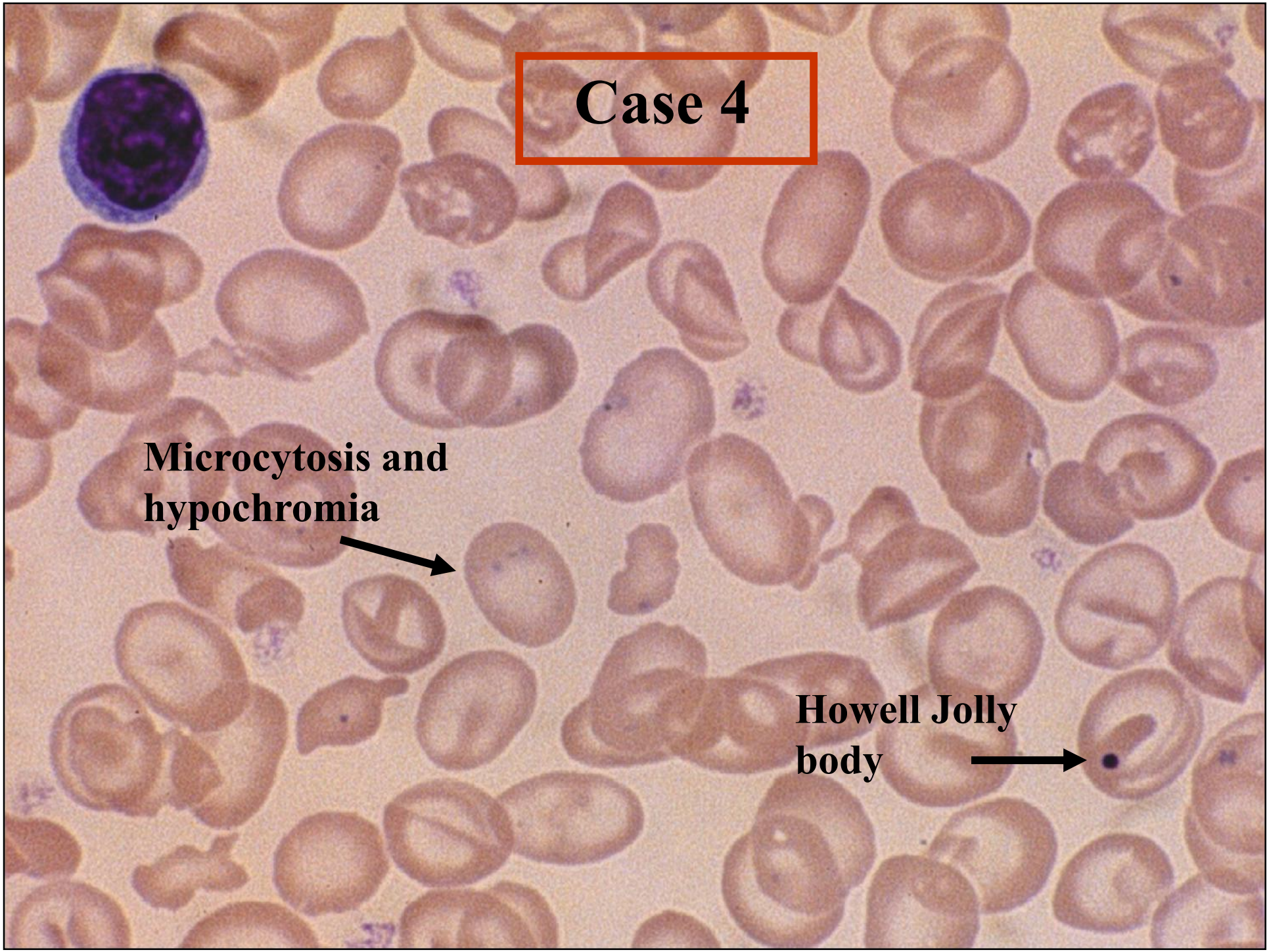
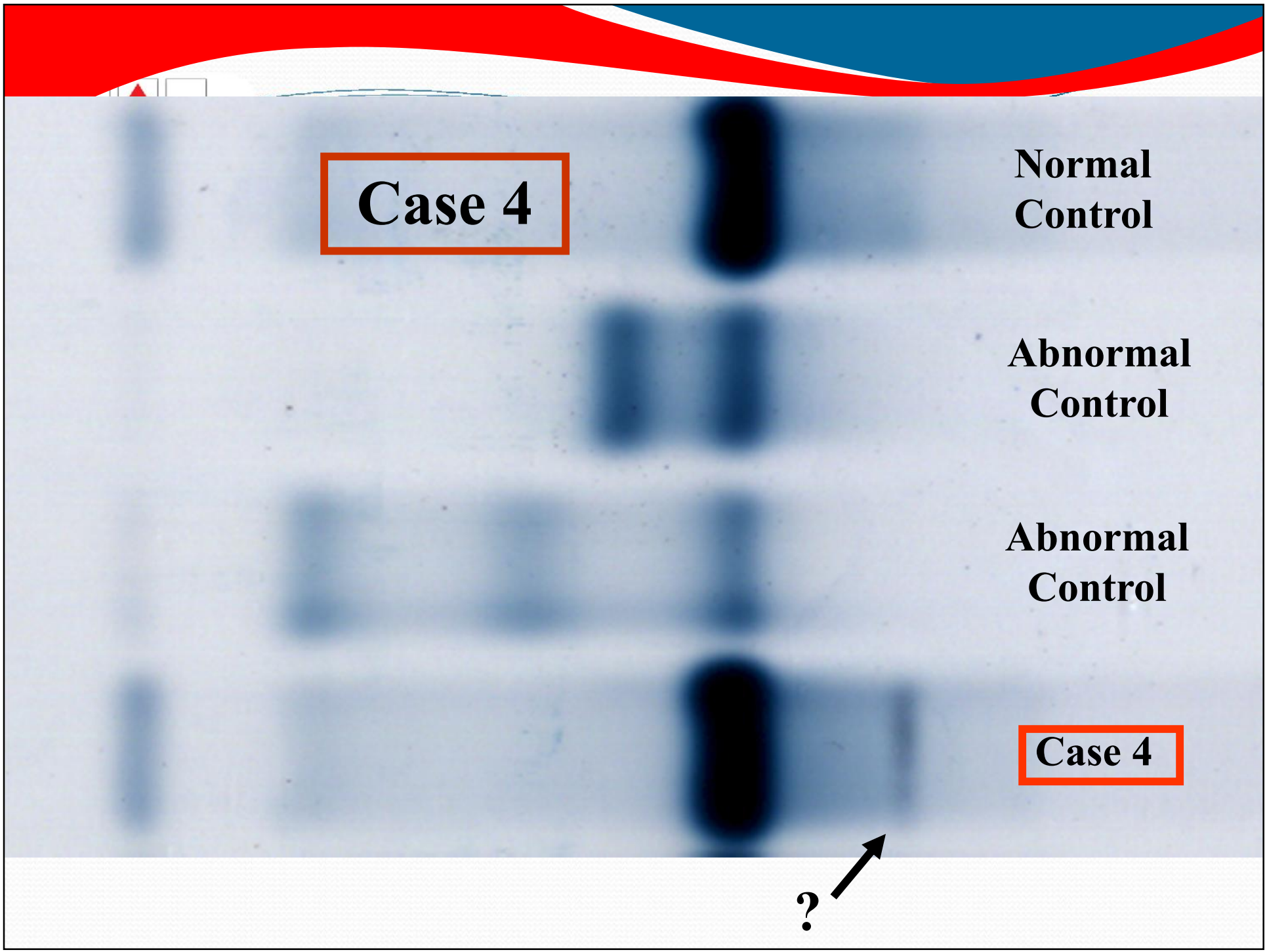
Case 4

Hb	**97 g/L (140-180)**
MCV	**72.1 fl (80-100)**


Case 4
Microcytosis and hypochromia
Howell Jolly body


Case 4
Normal Control
Abnormal Control
Abnormal Control
Case 4
?

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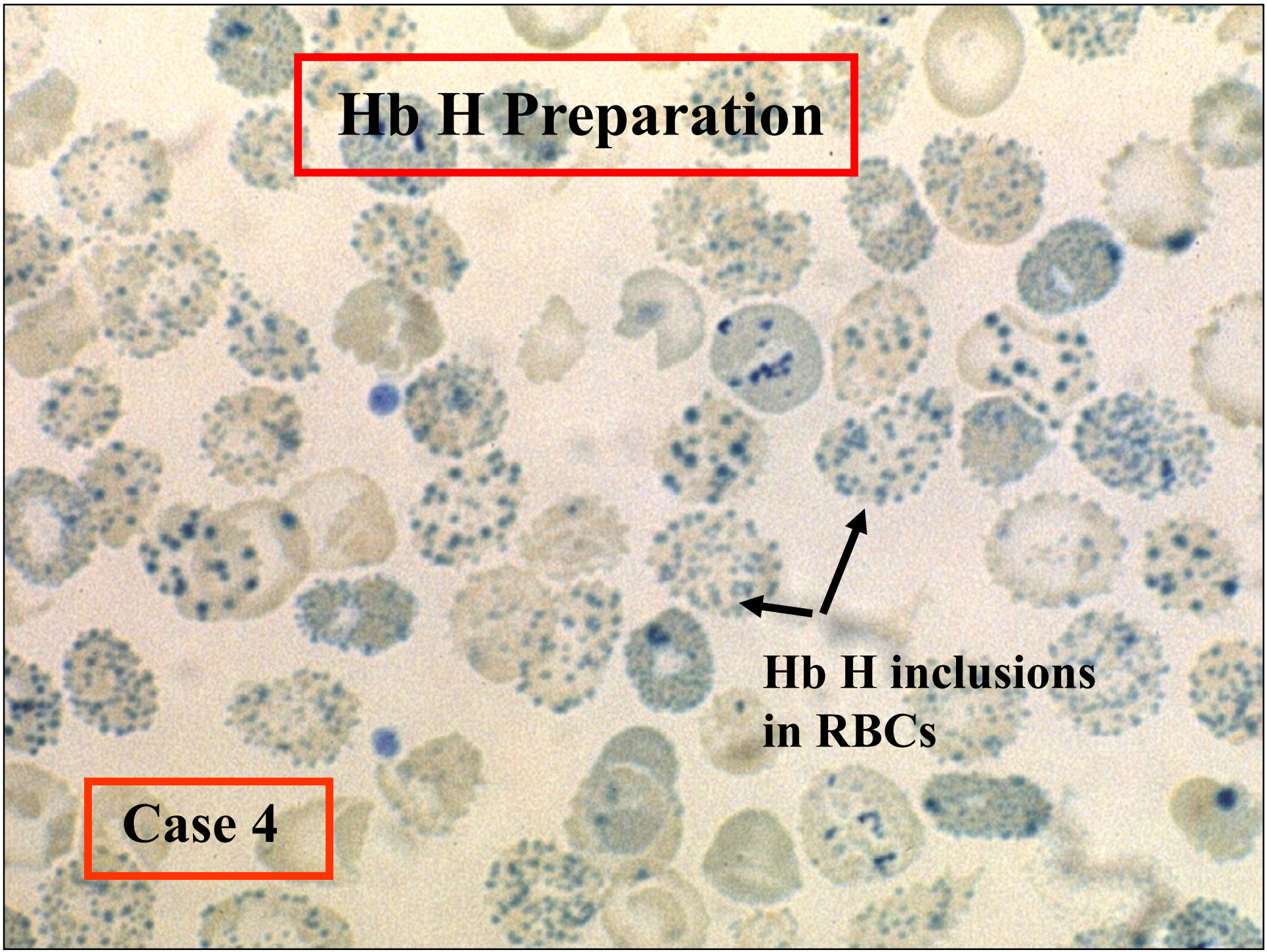
Case 4

Haemoglobin Electrophoresis:

Hb A 91%

Fast moving band 8.5 %

Hb A_2 and Hb F < 1%

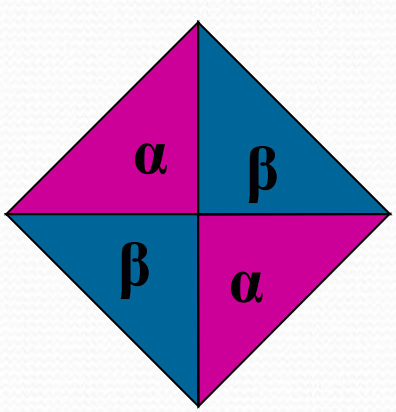

Hb H Preparation
Hb H inclusions
in RBCs
Case 4

Hb H Disease

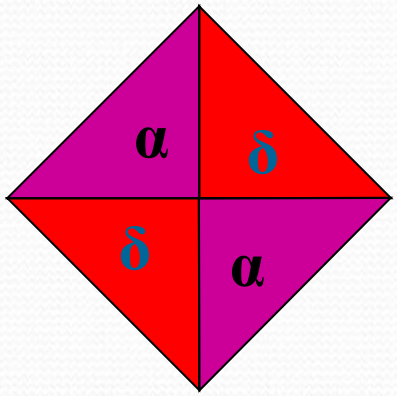
Diagnosis: Hb H Disease

Genotype -αβ/--β

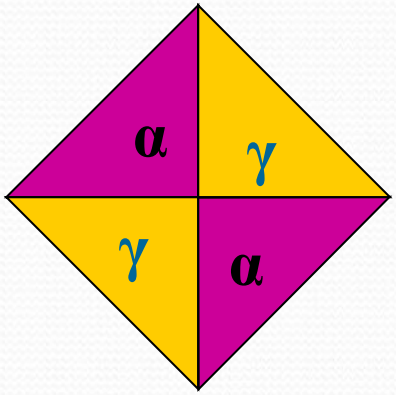
Haemoglobins Produced :



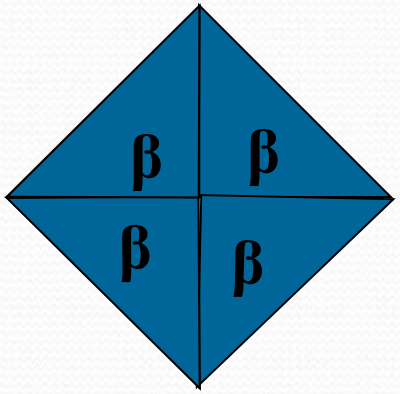
HbA



Hb A2



Hb F



Hb H