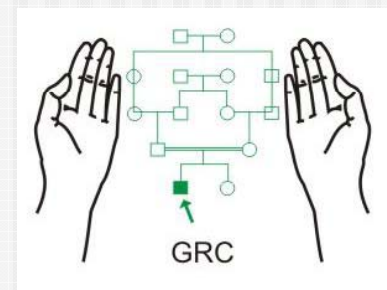


Screening Methods for β -Thalassaemia Trait

Maj Gen (R) Suhaib Ahmed, HI (M)

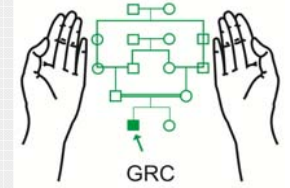
MBBS; MCPS; FCPS (Pak); PhD (London)

Genetics Resource Centre (GRC)



www.grcpk.com

Investigations for a Genetic Haemoglobin Disorder



Complete Blood Counts



Haemoglobin Electrophoresis



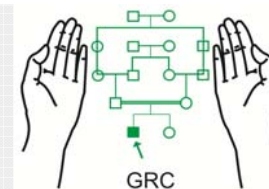
Quantitative Estimation of Hb Fractions



Other related Investigations



PCR



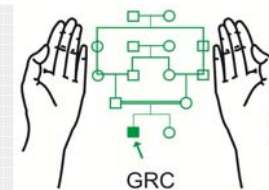
Blood Counts

No. 28
DATE: 22/ 3/95
MODE: WHOLE BLOOD

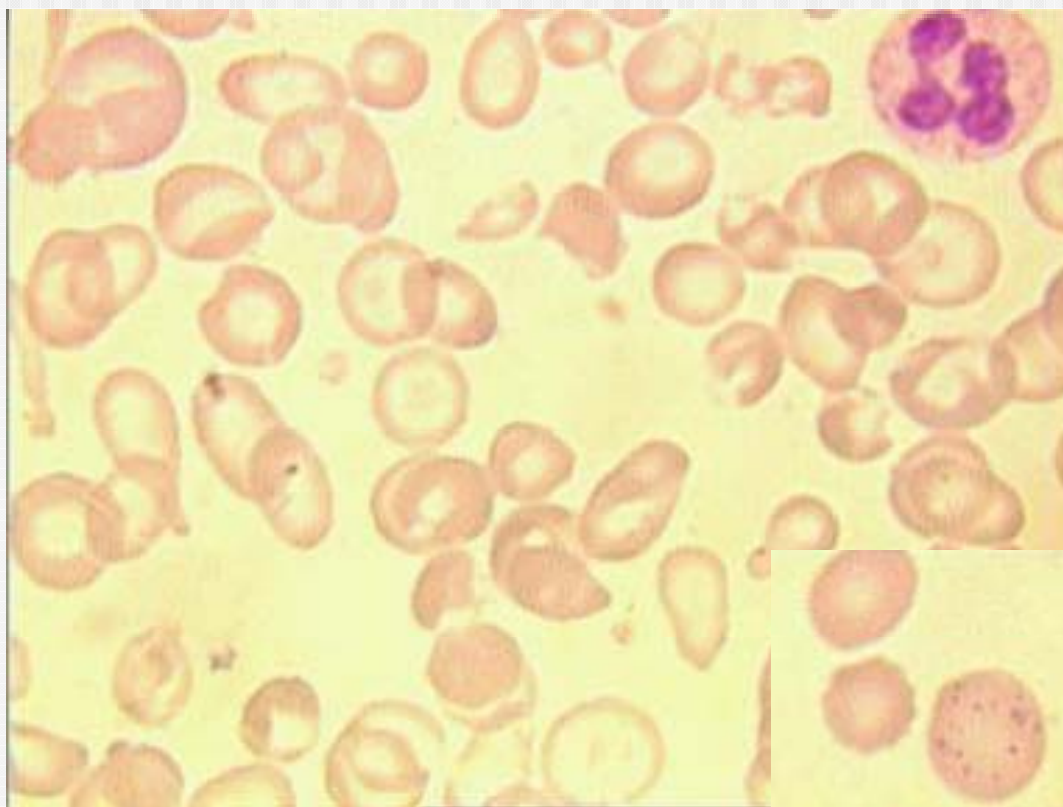
WBC	6.7	$\times 10^3 / \mu l$
RBC	5.45	$\times 10^6 / \mu l$
HGB	- 10.5	g/dl
HCT	- 33.4	%
MCV	- 61.3	fL
MCH	- 19.3	pg
MCHC	31.4	g/dl
PLT	318	$\times 10^3 / \mu l$

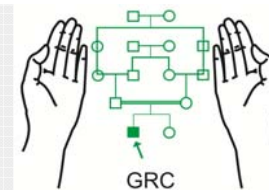
No. 32
DATE: 22/ 3/95
MODE: WHOLE BLOOD

WBC	8.9	$\times 10^3 / \mu l$
RBC	4.86	$\times 10^6 / \mu l$
HGB	14.4	g/dl
HCT	41.8	%
MCV	86.0	fL
MCH	29.6	pg
MCHC	34.4	g/dl
PLT	271	$\times 10^3 / \mu l$



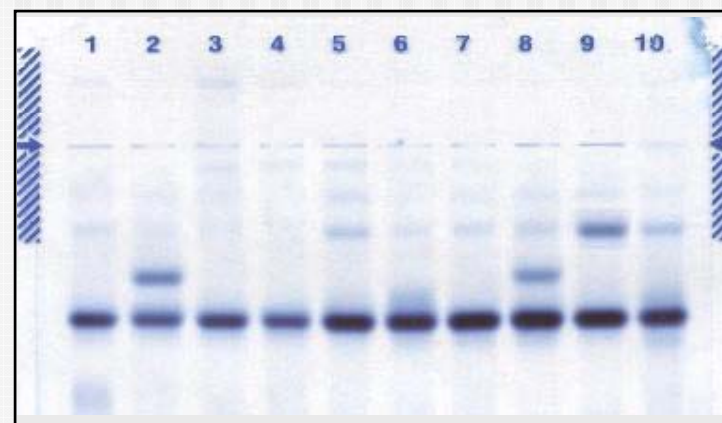
Peripheral Blood Film



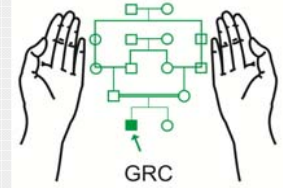


Hb-Electrophoresis

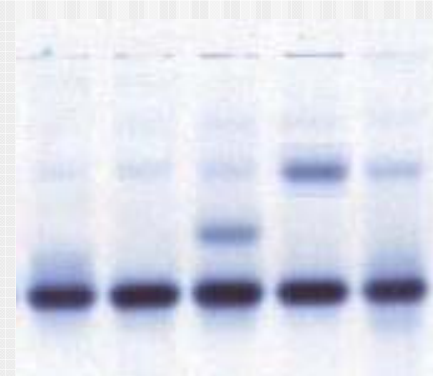
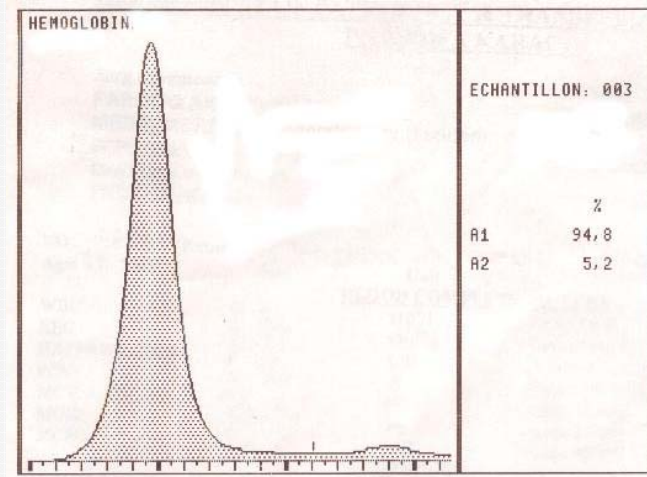
- Cellulose Acetate Membrane
- Cellogel
- Agarose Gel
- Agar Gel
- Polyacrylamide Gel

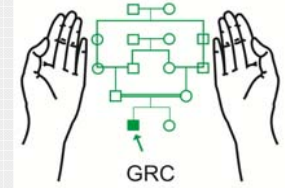


Quantitative Estimation of Hb Fractions



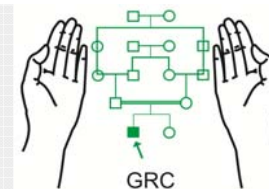
- How?
 - Chromatography
 - Elution after Electrophoresis
 - Densitometry??
- Where?
 - Hb-A₂
 - Others??



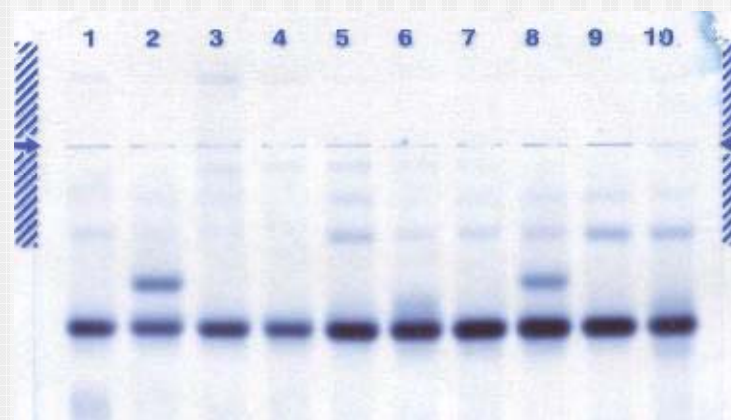
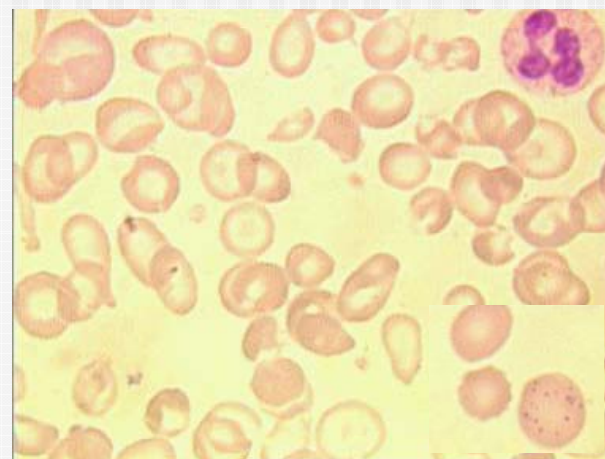
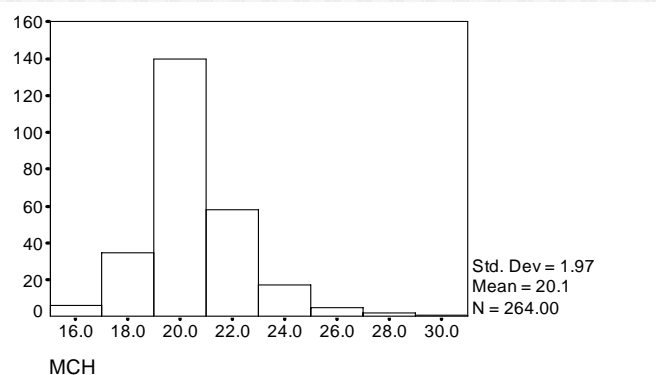
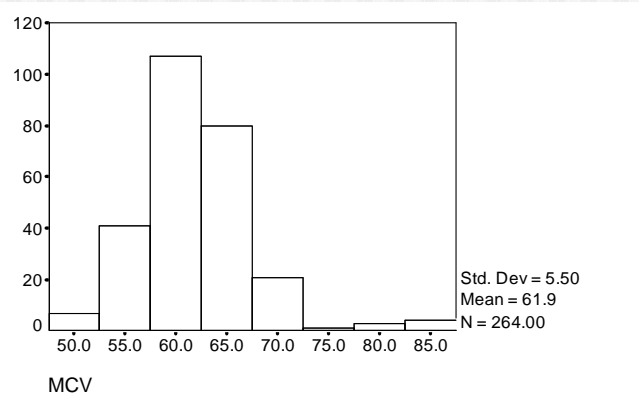


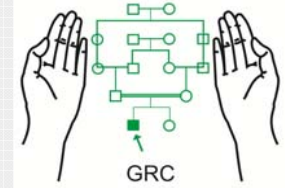
β -Thalassaemia Trait

- Typical form
- Silent β -thalassaemia trait
- Interaction with α -thalassaemia
- Interaction with structural Hb variants

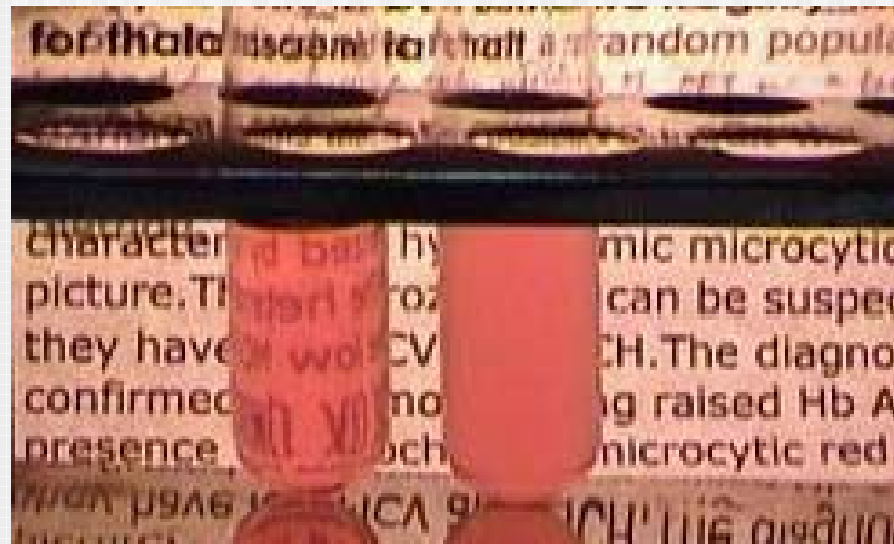


Typical β -thalassaemia Trait

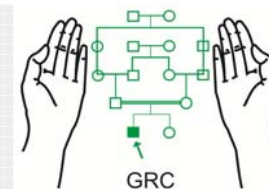




One Tube Osmotic Fragility Test



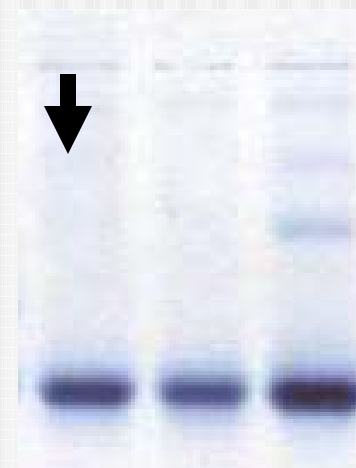
0.36% Saline



Silent β -thalassaemia Trait

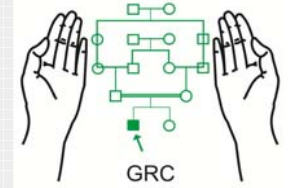
No. 58
DATE: 22/ 3/95
MODE: WHOLE BLOOD

WBC	9.2	$\times 10^9/\mu\text{l}$
RBC	4.36	$\times 10^6/\mu\text{l}$
HGB	- 11.7	g/dl
HCT	- 35.0	%
MCV	80.3	fL
MCH	26.8	pg
MCHC	33.4	g/dl
PLT	368	$\times 10^3/\mu\text{l}$

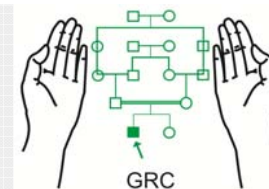


PCR: Cap+1 or -88 mutation
(2% of β -thalassaemia carriers)

Co-existing α and β -thalassaemia Trait



- Approximately 2% of β -thalassaemia carriers have co-existing α -thalassaemia trait
- Red cell indices are normalized
- Hb-A₂ is increased
- PCR may be required



β -Thalassaemia Trait and Interacting Structural Hb Variants

No.	82	
Date	24/ 6/02 13:01	
Mode	Whole Blood	
WBC	7.2	$\times 10^3/\mu\text{L}$
RBC	5.88	$\times 10^6/\mu\text{L}$
HGB	9.9	g/dL
HCT	33.3	%
MCV	56.6	fL
MCH	16.8	pg
MCHC	29.7	g/dL
PLT	306	$\times 10^3/\mu\text{L}$



Hb-D/ β -thalassaemia Trait

Screening Strategy for Thalassaemia Carriers

