

IN THE NAME OF ALLAH, THE MOST BENEFICIENT, THE MOST MERCIFUL

**FREQUENCY OF BETA THALASSAEMIA TRAIT IN INDIVIDUALS
WITH AND WITHOUT
ISCHAEMIC HEART DISEASE**

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β-THALASSAEMIA

HISTORICAL BACKGROUND

- Dr Thomas B. Cooley (1925)
- Whipple and Bradford (1936)
- The word “Thalassaemia” means the “the sea” in Greek

Postgraduate haematology, fifth edition

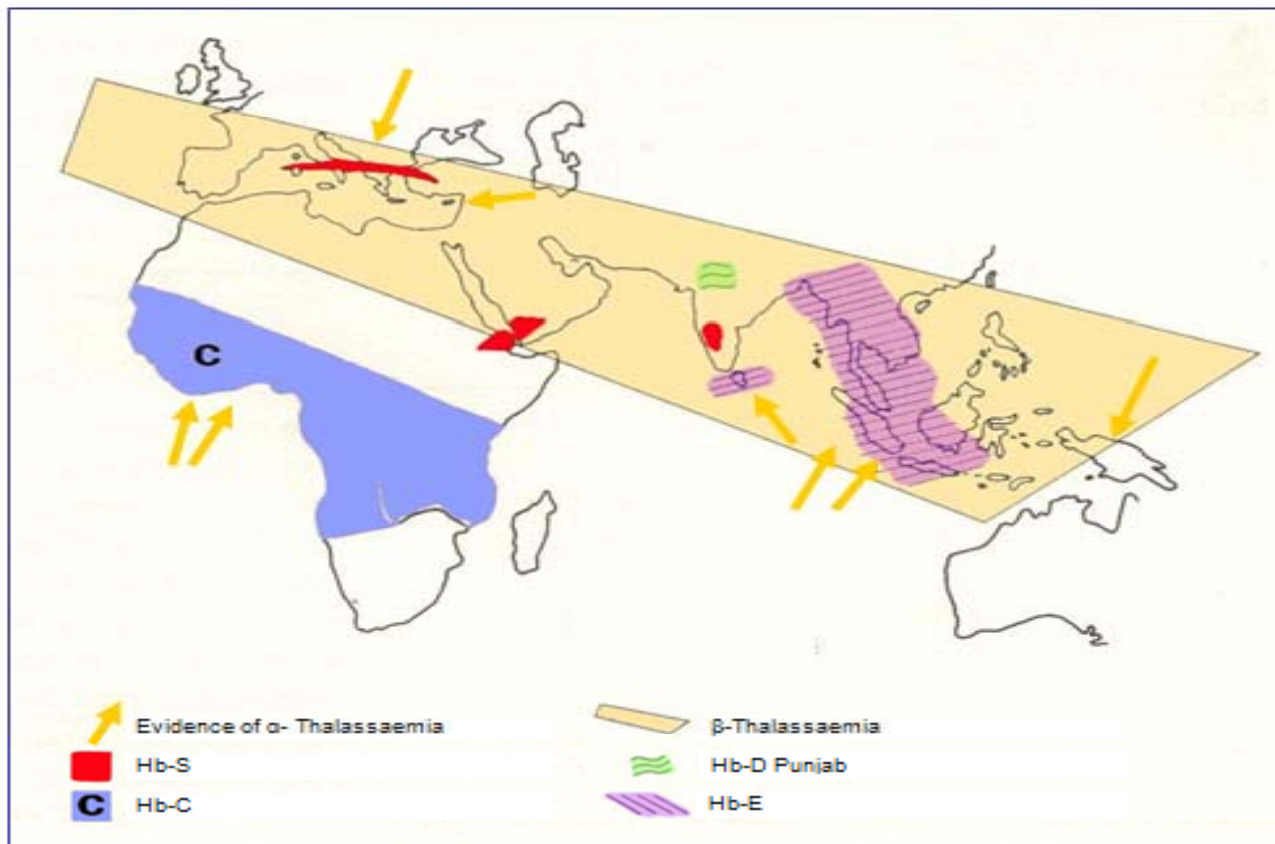
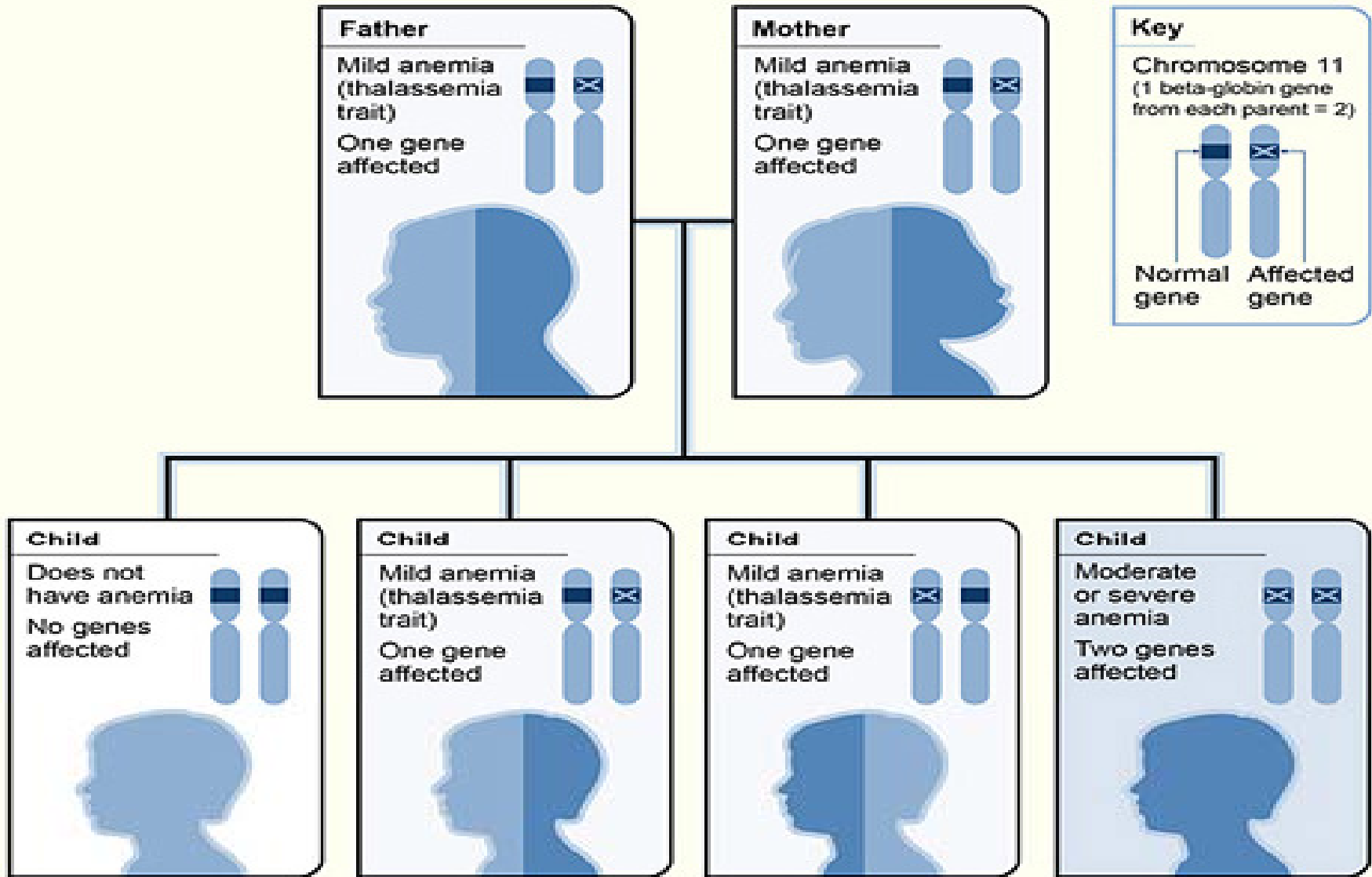
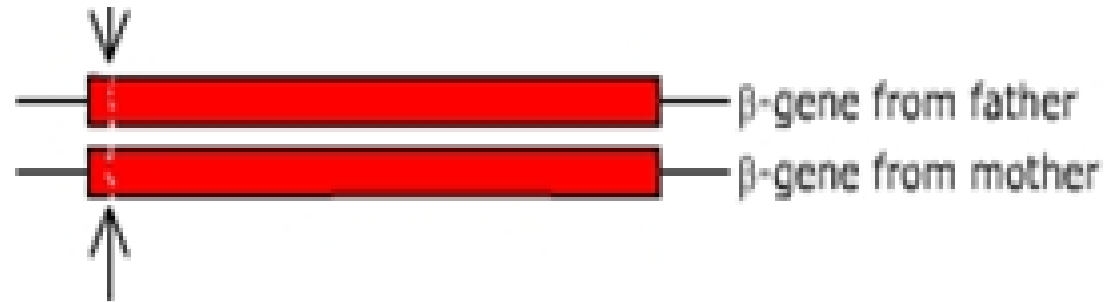


FIGURE 1: Geographical distribution of hemoglobin disorders



With a mutation on one of the two β -globin genes, a carrier is formed with lower protein production, but enough hemoglobin



**Without a mutation
enough Hemoglobin**



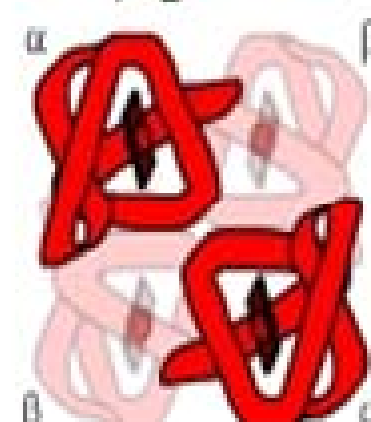
No thalassemia carrier

**With one mutation
less Hemoglobin**

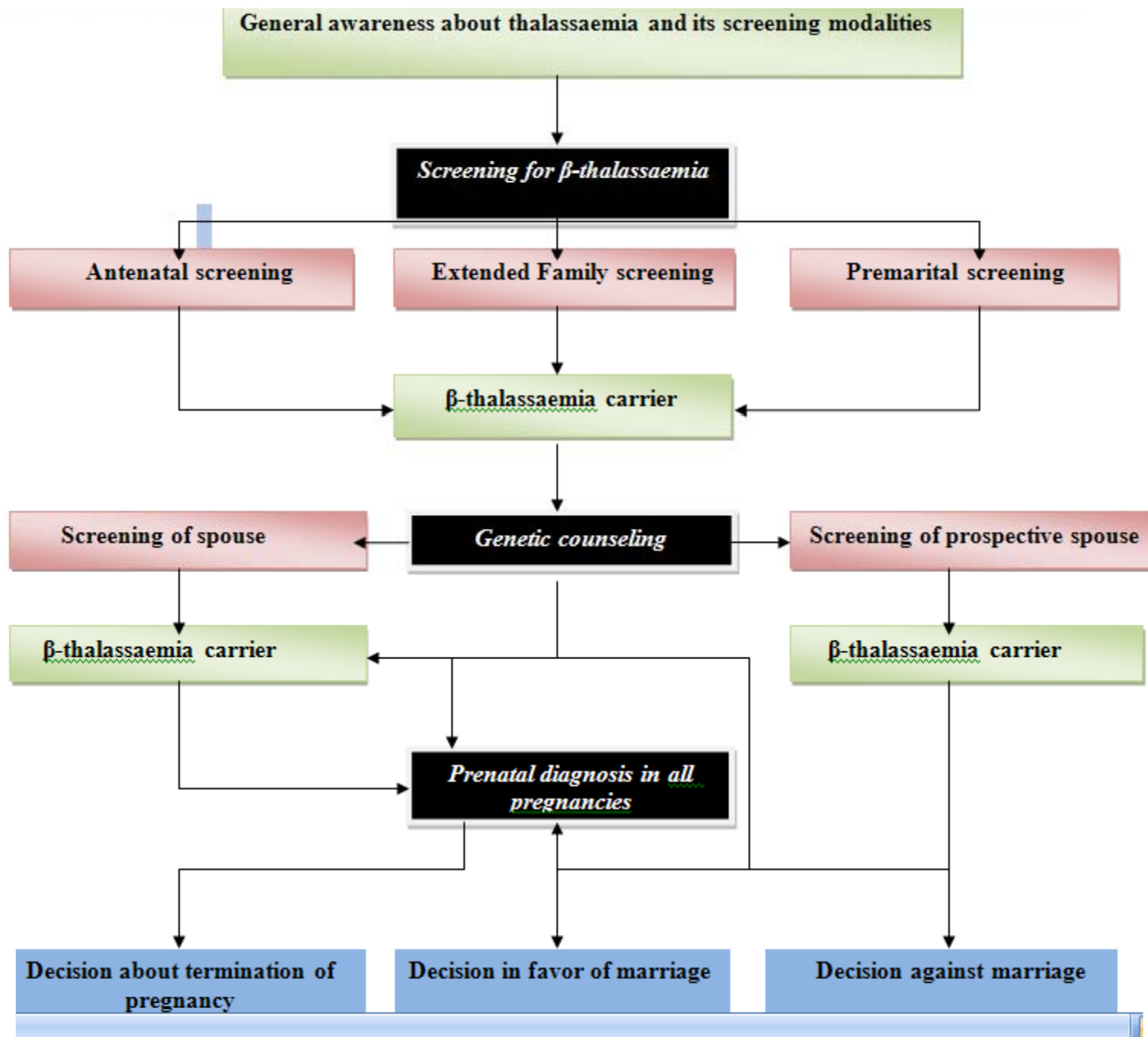


β -thalassemia carrier without illness, but less hemoglobin (slight anemia)

**With two mutations
no β -globin**



β -thalassemia major patient with severe anemia





The New England Journal of Medicine

SPECIAL ARTICLE
2002; 347: 1162-1168

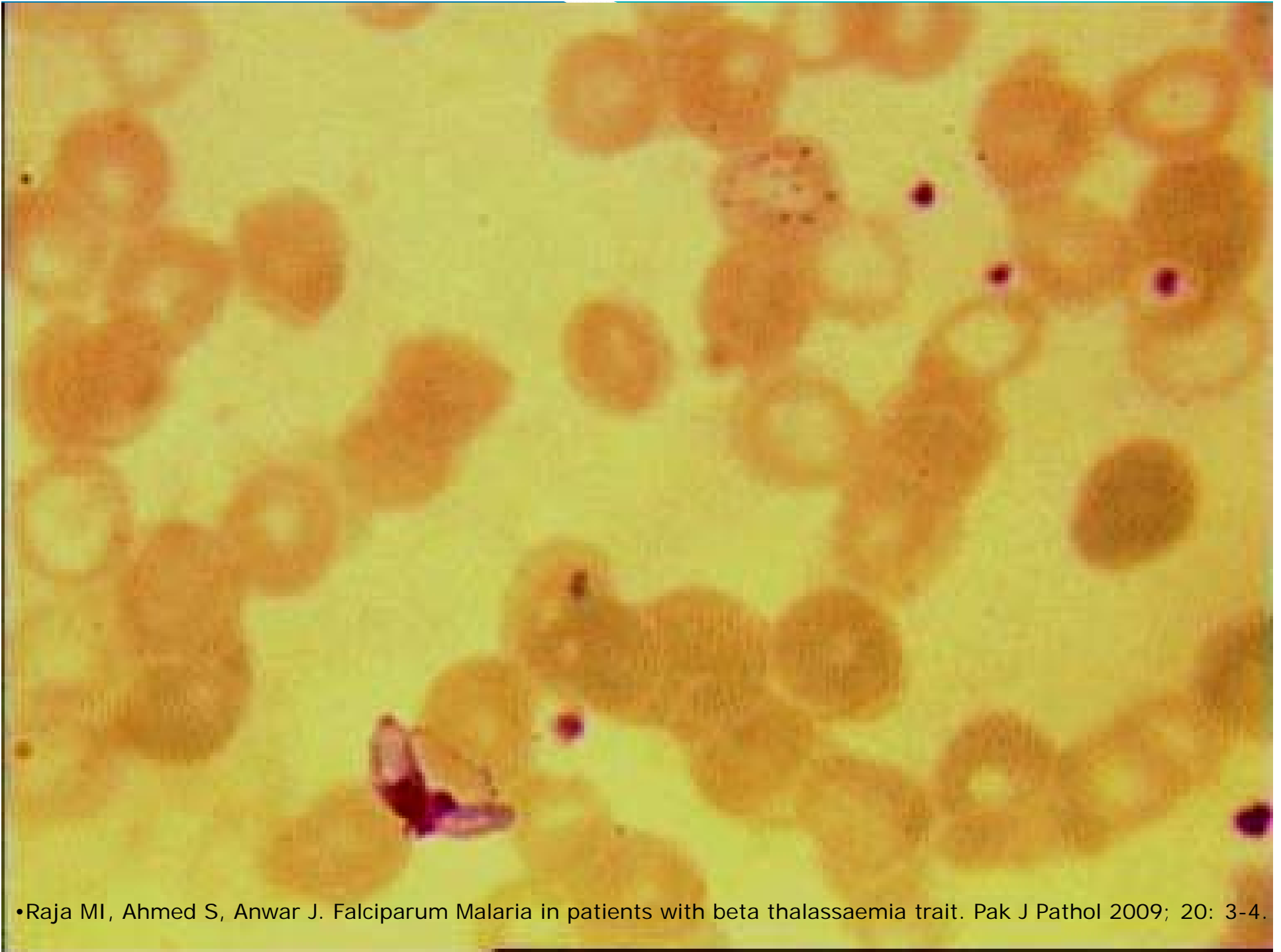
Screening Extended Families for Genetic Haemoglobin Disorders in Pakistan.

Suhaib Ahmed; Mohammad Saleem; Bernadette Modell;
Mary Petrou.



GENETIC COUNSELING

- WHAT IS GENETIC COUNSELING?
- WHAT NEEDS TO BE EXPLAINED?
 - Marriage
 - Screening
 - Prenatal diagnosis
- HOW TO CURB THE NEGATIVE FEELINGS?
 - Sharing the possible benefits associated with the disorder



•Raja MI, Ahmed S, Anwar J. Falciparum Malaria in patients with beta thalassaemia trait. Pak J Pathol 2009; 20: 3-4.

PROTECTION FROM ISCHAEMIC HEART DISEASE

- **INTERNATIONAL STUDIES VALIDATING THE HYPOTHESIS**

- Tassiopoulos S, Deftereos S, Konstantopoulos K, Farmakis D, Tsironi M, Kyriakidis M, et al. Does heterogenous beta thalassaemia confer a protection against coronary artery disease? *Ann N Y Acad Sci* 2005; 1054: 467-70.
- Wang CH, Shilling RF. Myocardial infarction and thalassemia trait: an example of heterozygote advantage. *Am J Haematol* 1995; 49: 73-5

- **LOCAL STUDIES**



OBJECTIVE

- To compare the frequency of beta thalassaemia trait in patients of Ischaemic heart disease and a control population without Ischaemic heart disease



HYPOTHESIS

- Frequency of beta thalassaemia trait is low in IHD patients than in normal population

Tassiopoulos S, Deftereos S, Konstantopoulos K, Farmakis D, Tsironi M, Kyriakidis M, et al. Does heterogenous beta thalassaemia confer a protection against coronary artery disease? *Ann N Y Acad Sci* 2005; 1054: 467-70



MATERIAL AND METHODS

- **STUDY DESIGN**

- Case control study

- **SETTING**

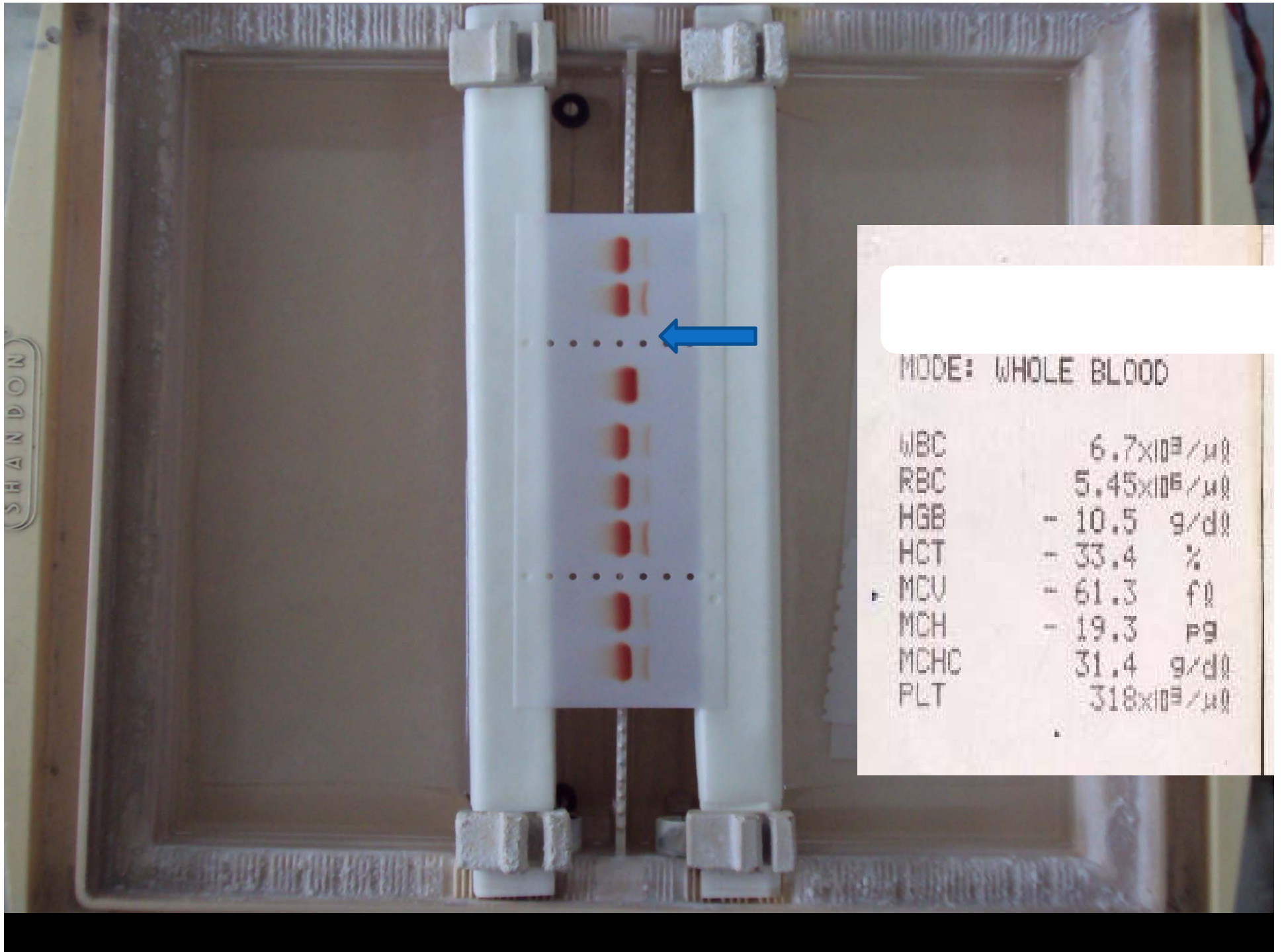
- Department of Haematology, Armed Forces Institute of Pathology, Rawalpindi

- **DURATION**

- September 2007 to May 2009

- **SAMPLE SIZE**

- n=544



MODE: WHOLE BLOOD

WBC	6.7	$\times 10^9/\mu\text{l}$
RBC	5.45	$\times 10^6/\mu\text{l}$
HGB	- 10.5	g/dl
HCT	- 33.4	%
MCV	- 61.3	fL
MCH	- 19.3	pg
MCHC	31.4	g/dl
PLT	318	$\times 10^9/\mu\text{l}$



RESULTS

DEMOGRAPHICS

- Age
- Gender
- Ethnic distribution

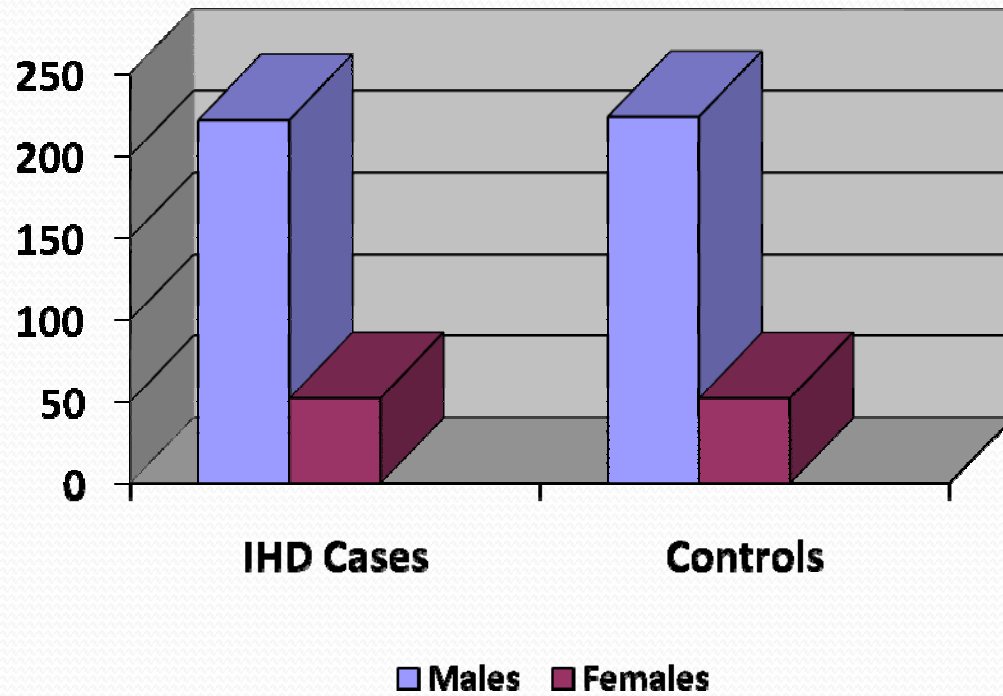
BLOOD COUNTS

HAEMOGLOBIN ELECTROPHORESIS

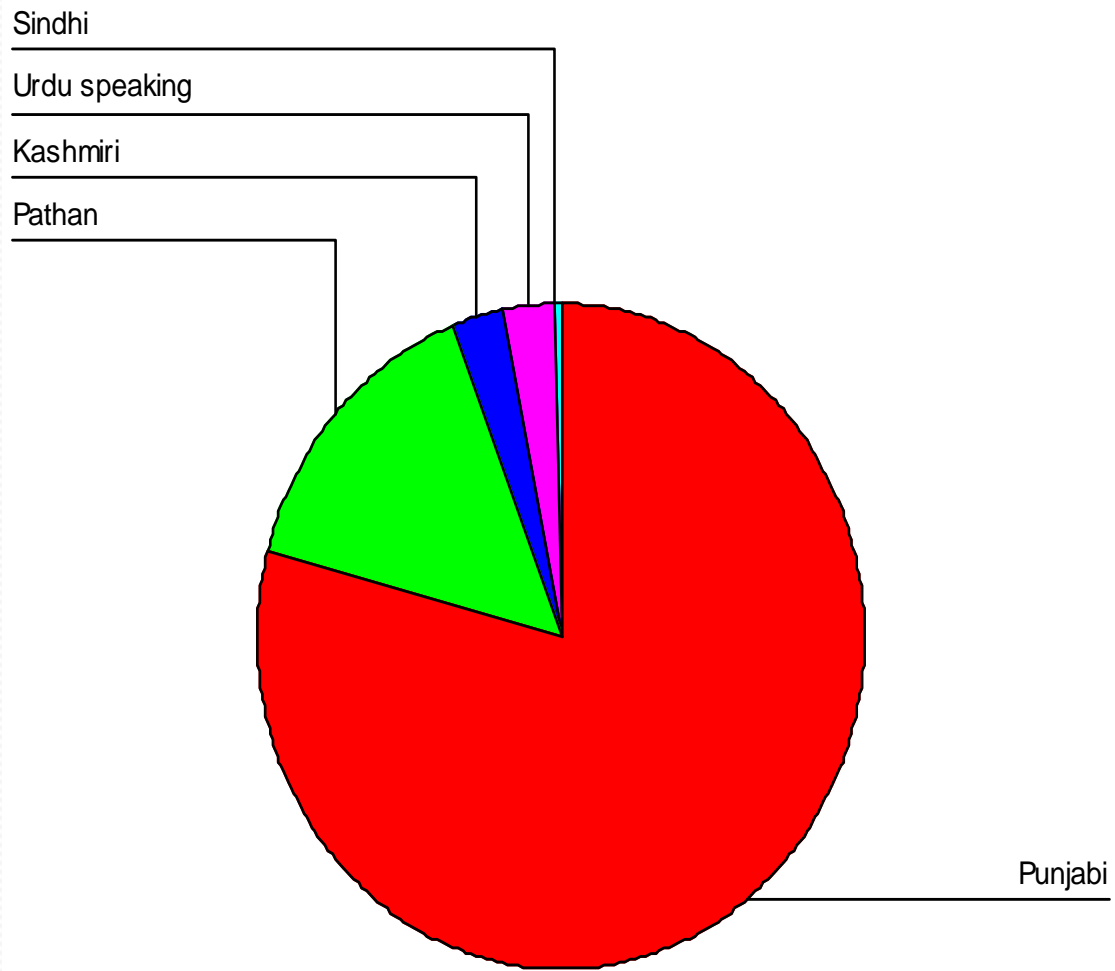
Summary of age statistics

Group	Minimum age	Maximum age	Mean age	Standard deviation	Median age
A	39	79	60	8.18	63
B	40	78	61	6.58	61.5
Total	39	79	60.5	7.38	62.25

- Group B: Normal Controls
- Group A: IHD Cases

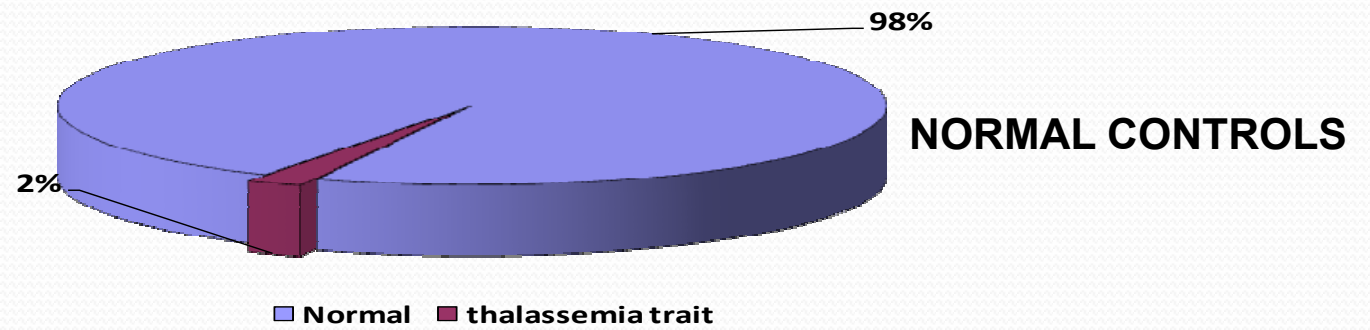
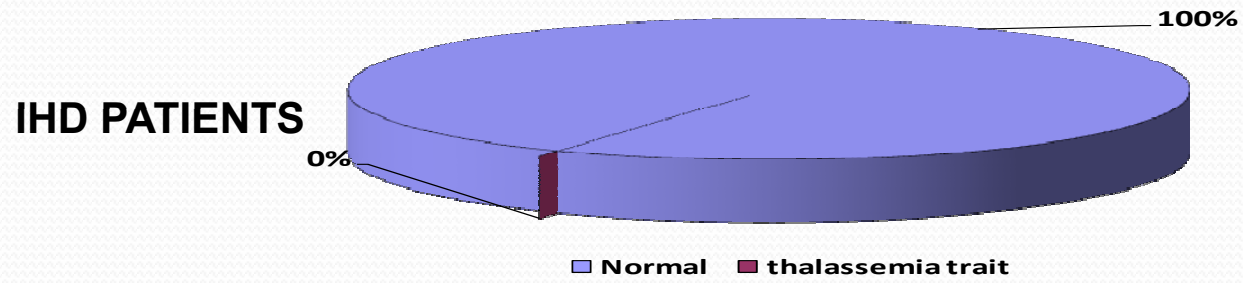


Gender distribution in both groups



Ethnic distribution of the study population

•n = 544



Frequency of β -thalassaemia trait in both groups

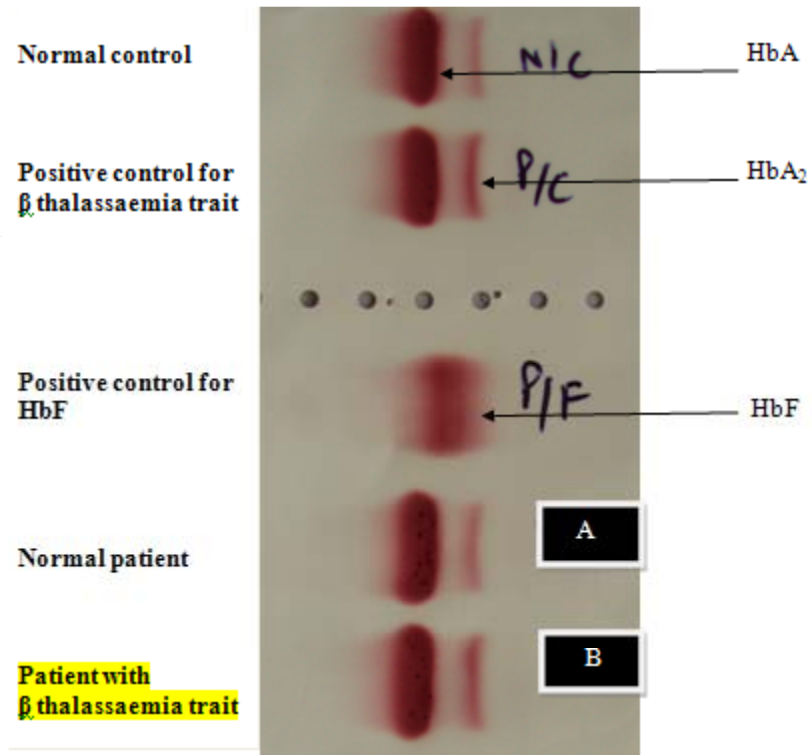


FIGURE 17: Electrophoresis strip showing a normal (A) a positive result (B) in the control group

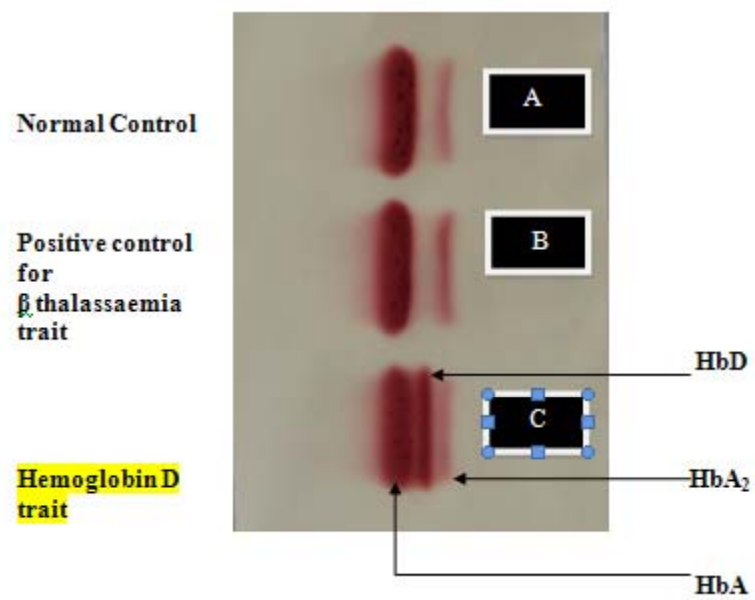


FIGURE 18: Electrophoresis strip showing a patient with Hemoglobin D Trait (C) in the control group

CALCULATION OF ODDS RATIO

	Disease		
	IHD (cases)	Non IHD	Total
β -thalassaemia trait	0 (A)	6 (B)	6
Non β -thalassaemia trait	272 (C)	266 (D)	538
Total	272	272	544
Odds Ratio (OR) = $A \times D / B \times C = 0 \times 266 / 6 \times 272 = 0$			

Odds ratio of < 1 shows non significant association

Where A = IHD patients with β -thalassaemia trait

B = Non IHD controls with β -thalassaemia trait

C = IHD patients with no β -thalassaemia trait

D = Non IHD controls with no β -thalassaemia trait

COMPARISON IN THE FREQUENCY OF β -THALASSAEMIA TRAIT IN THE TWO GROUPS

Group	β -thalassaemia trait		Total
	Yes	No	
A	6 (2.2%)	266 (97.8%)	272
B	0 (0%)	272 (100%)	272
total	6 (1.1%)	538 (98.9%)	544

- P value is 0.033, ($p = < 0.05$) thus the observed difference is statistically significant
- Group A: Normal control population
 - Group B: IHD patients



CONCLUSION

- Frequency of Beta thalassaemia trait is lower in IHD patients
- Beta Thalassaemia trait confers some protection against IHD
- The protection is not absolute due to role of other risk factors
- `Beneficial information in the counseling of beta thalassaemia carriers

A lit candle in a brass holder, with a small flame visible in the distance. The scene is set against a dark background, with the candle's light illuminating the holder and casting a warm glow. The text "Thank you" is written in red in the lower-left corner.

Thank you