

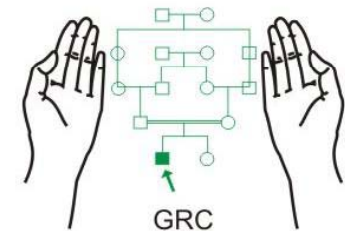
Extended Family Screening of Thalassaemia

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

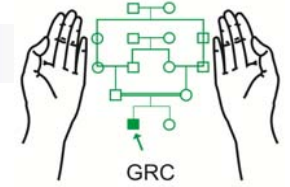
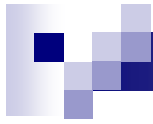
MBBS; MCPS; FCPS (Pak); PhD (London)

Genetics Resource Centre (GRC)

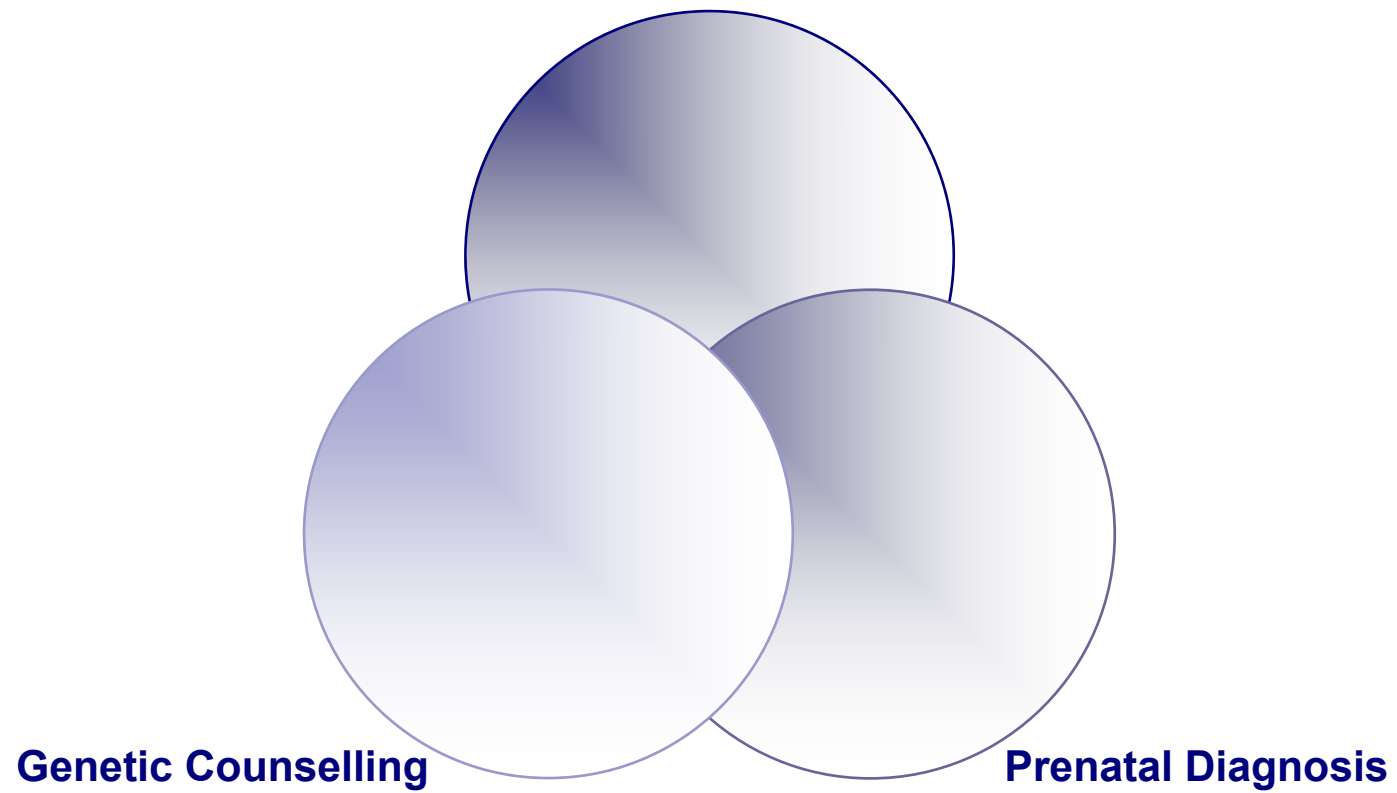
Rawalpindi

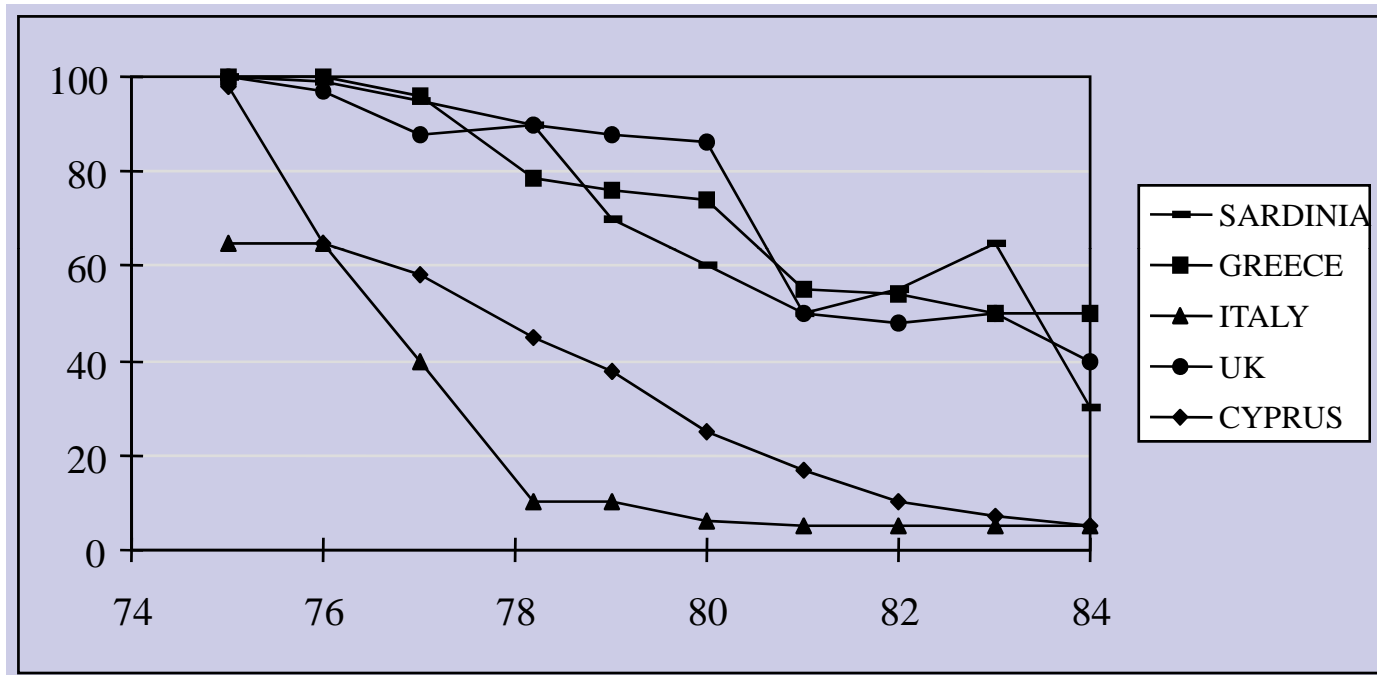
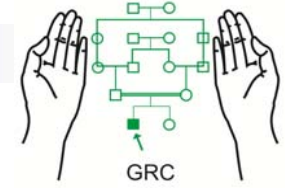


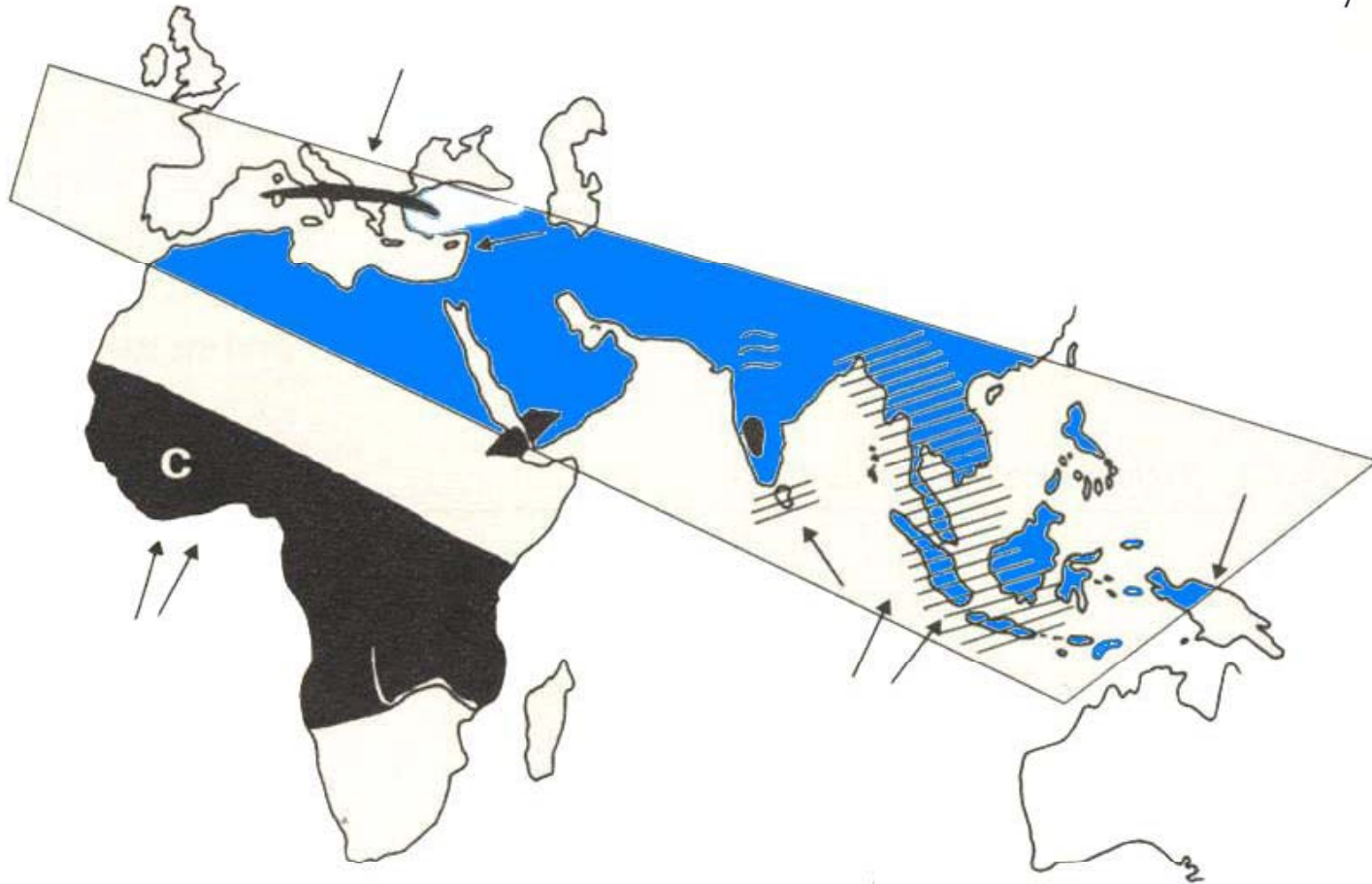
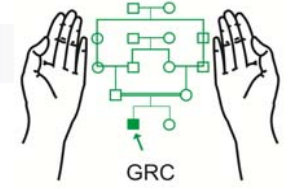
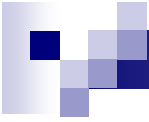
www.grcpk.com



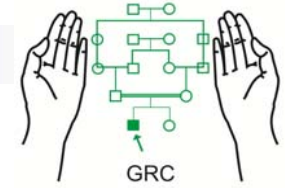
Carrier Screening





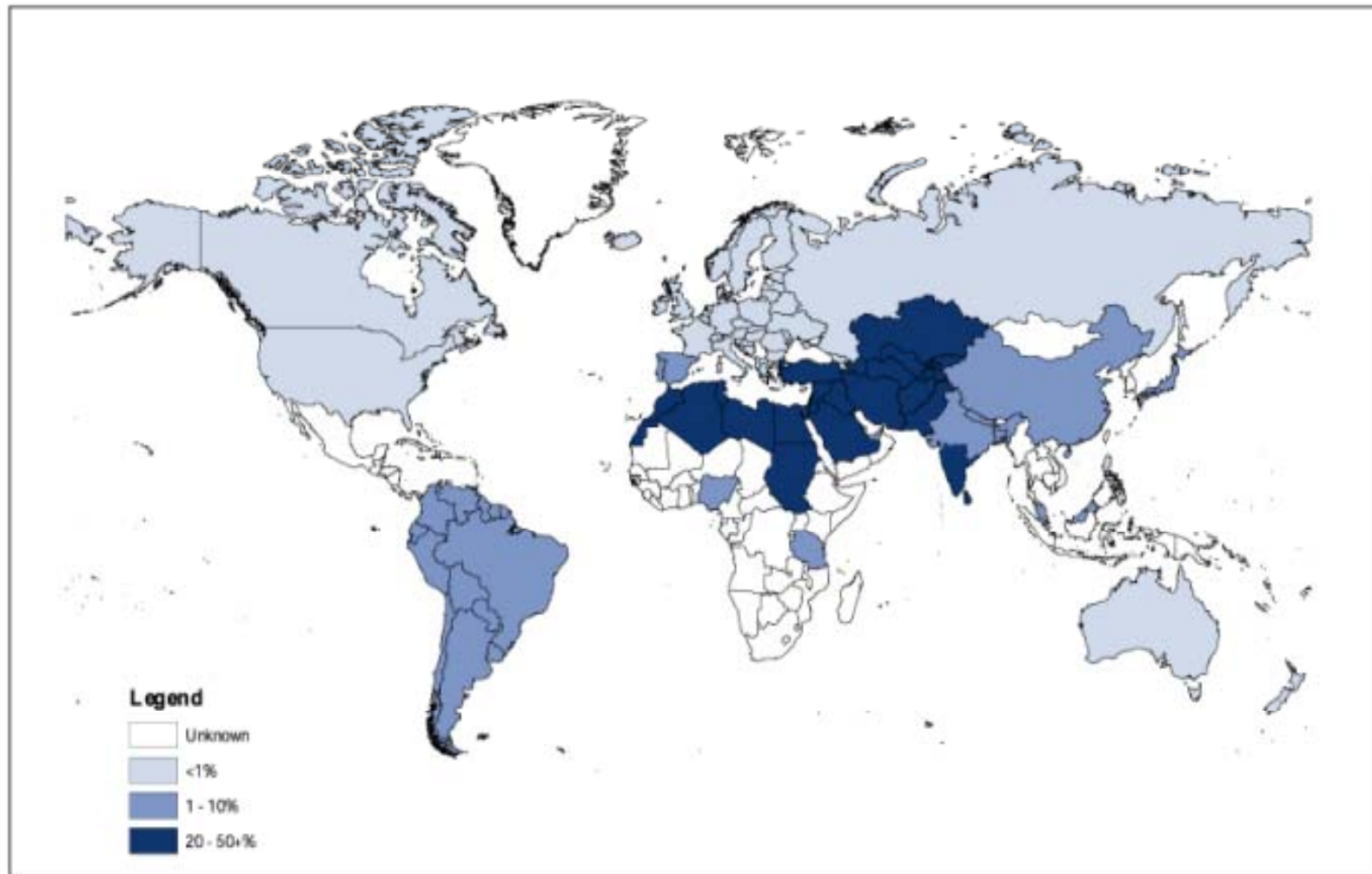
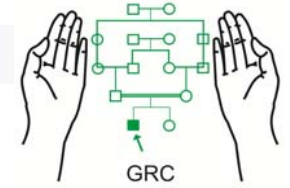


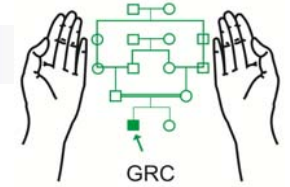
- ↗ Evidence for α -Thalassaemia
- Hb-S
- C Hb-C
- ▭ β -Thalassaemia
- ≡ Hb-D Punjab
- ▨ Hb-E



The Third World Scenario

- Lack of resources
- Disorganized resources
- Large population size
- Low level of literacy



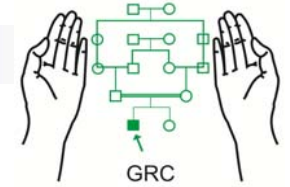


β -Thalassaemia Trait in Pakistan

- Punjabi 4.6 %
- Pathan 5.2 %
- Sindhi 4.3 %
- Baluchi 8.0 %
- Urdu speaking 5.3 %
- Overall 5.5 %



(Ahmed 1998)

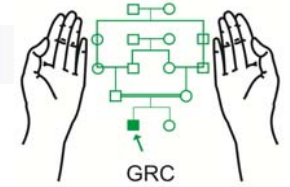


β -Thalassaemia Major in Pakistan

- Annual birth rate: 5000
- Total numbers: >50,000
- Registered cases: ~10,000
- Mean life expectancy: 10 years

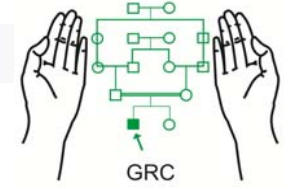


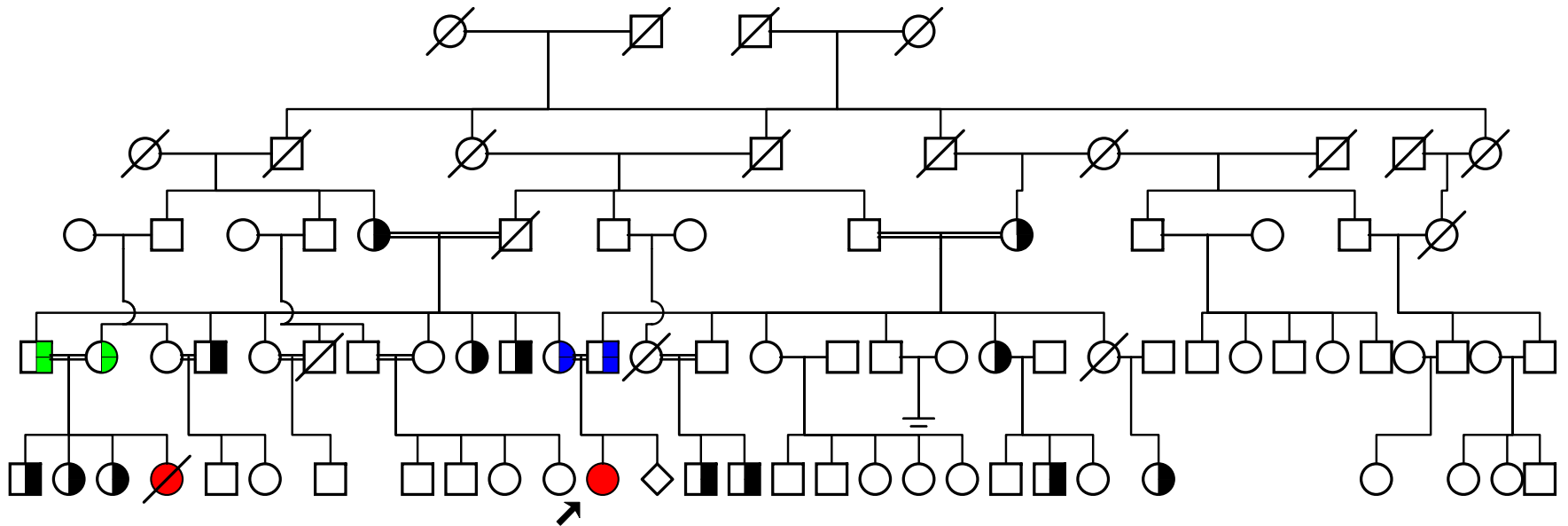
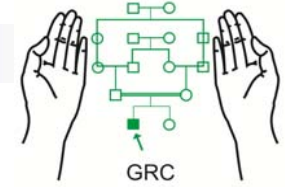
(Ahmed 1998)



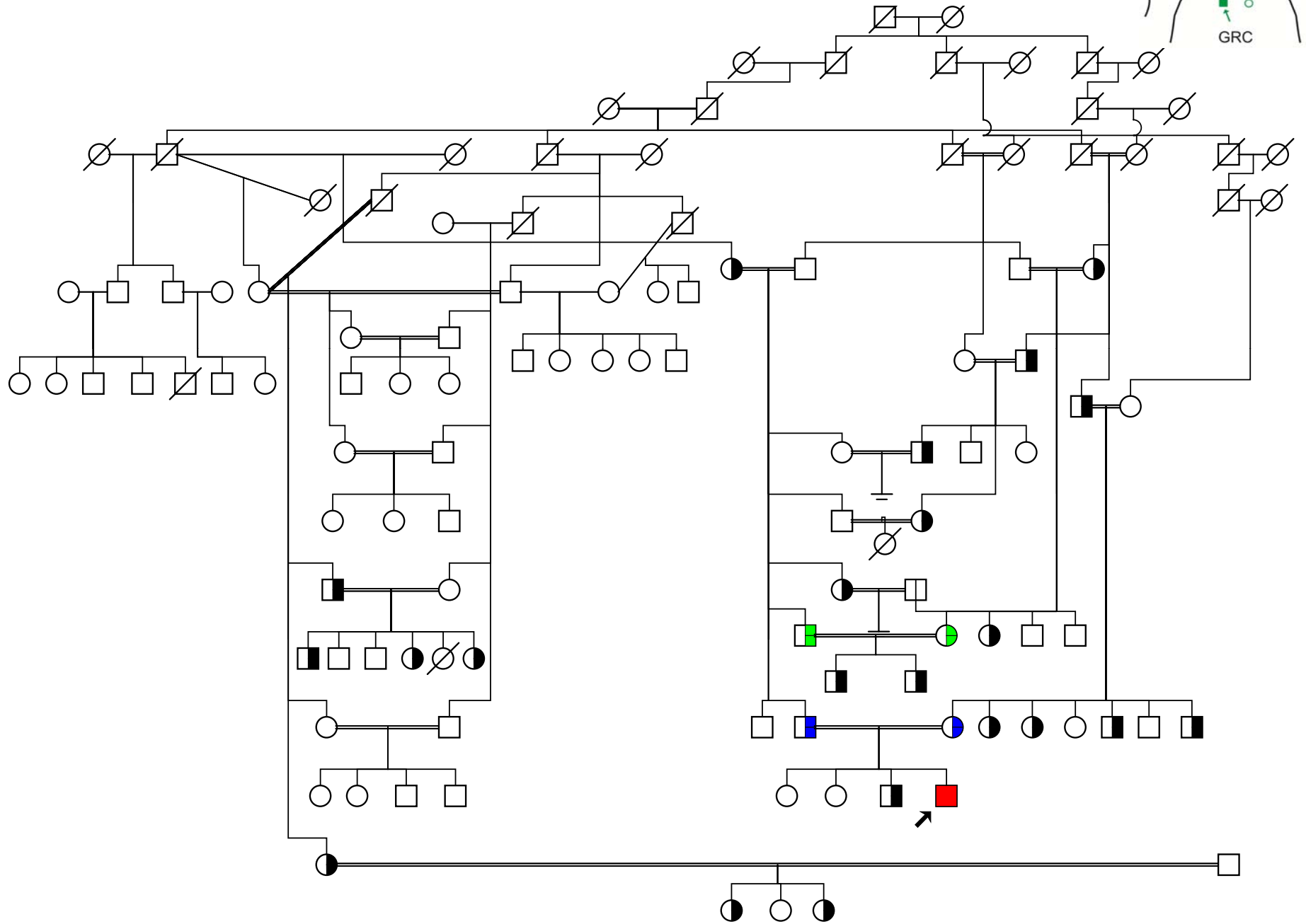
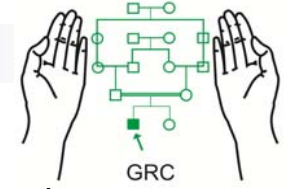
Carrier Screening

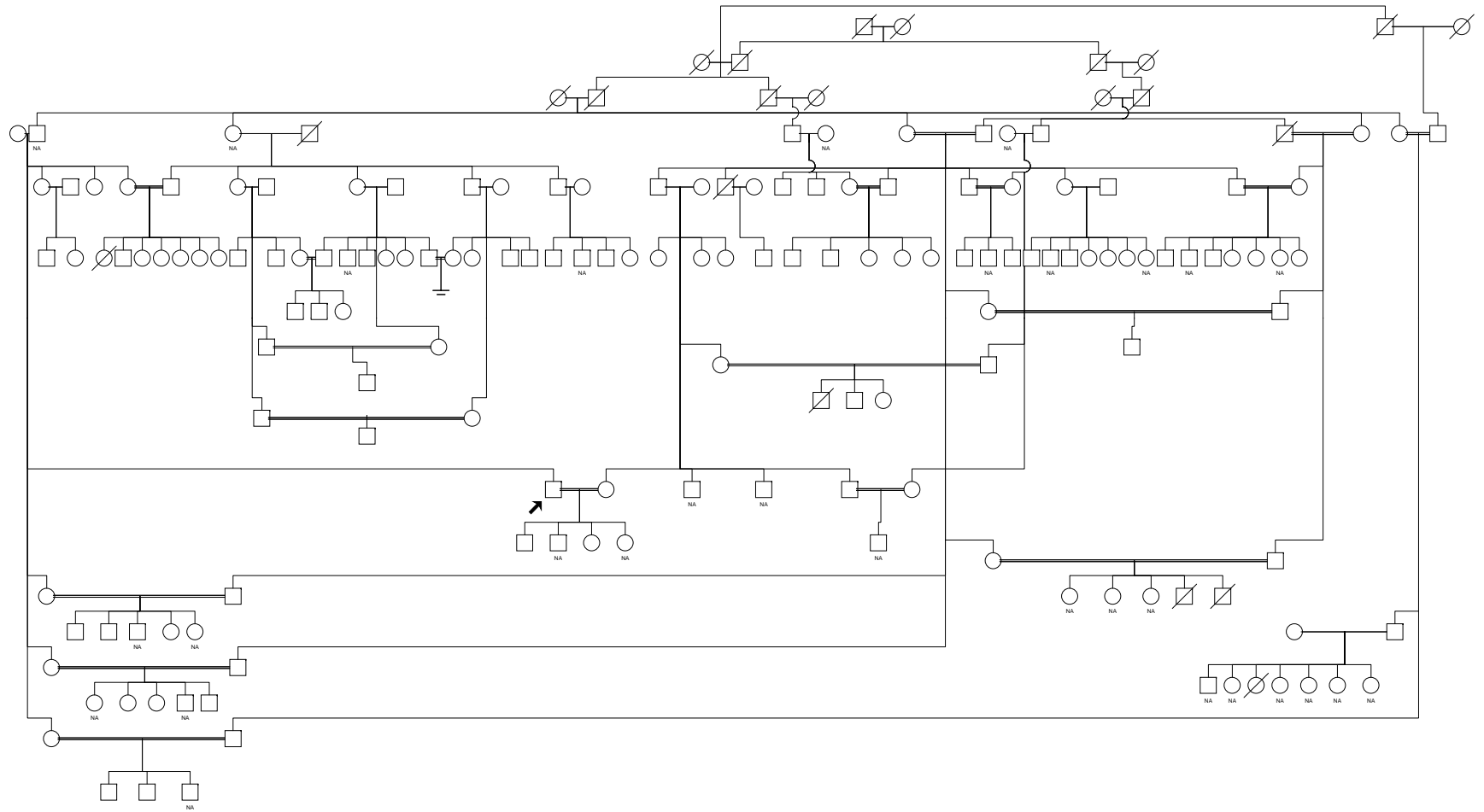
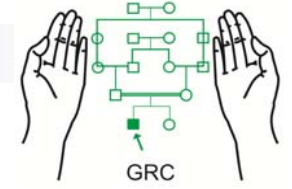
- Mass population screening
- Screening in Pregnancy
- **Extended Family Screening**

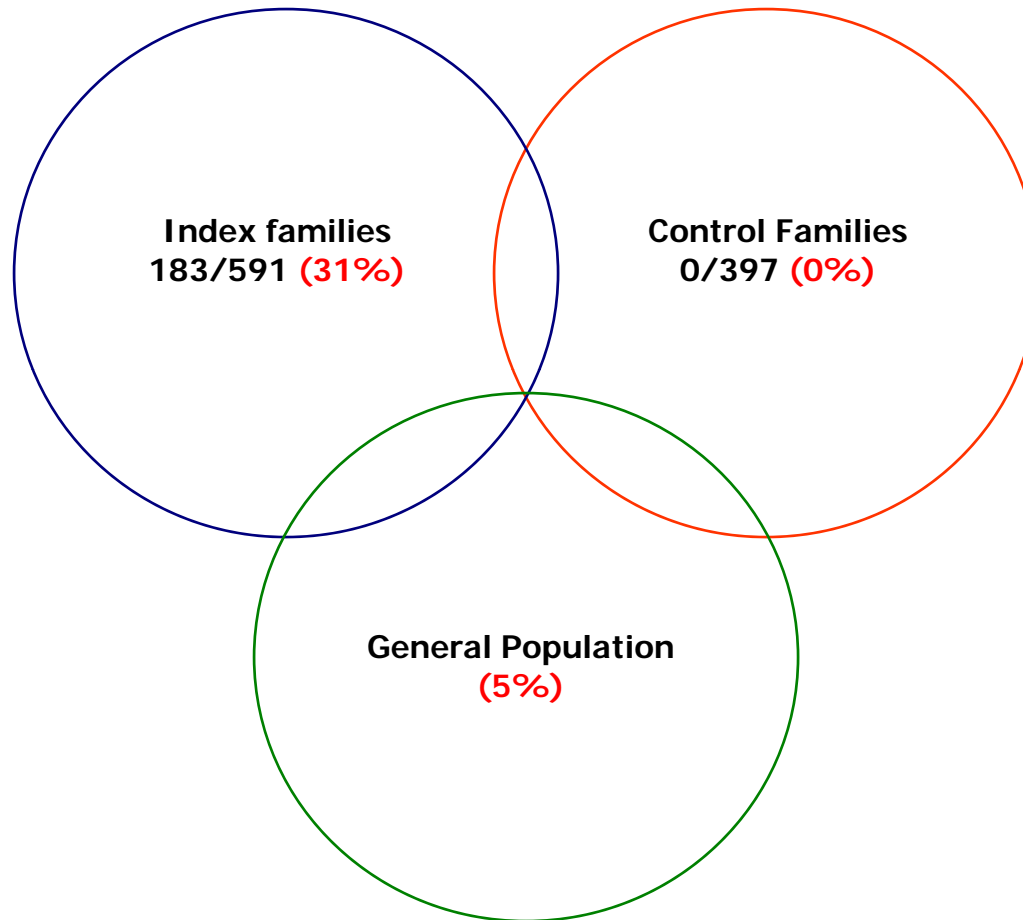
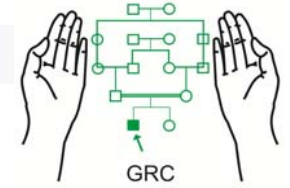


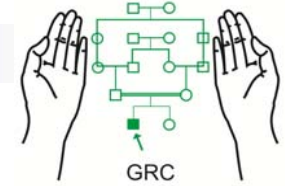


(Ahmed S. et al, 2002)



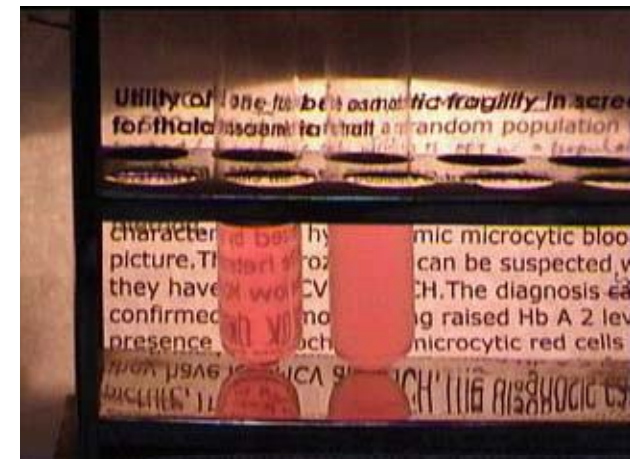


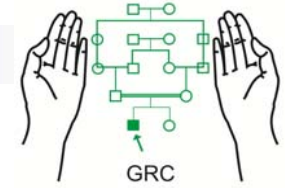




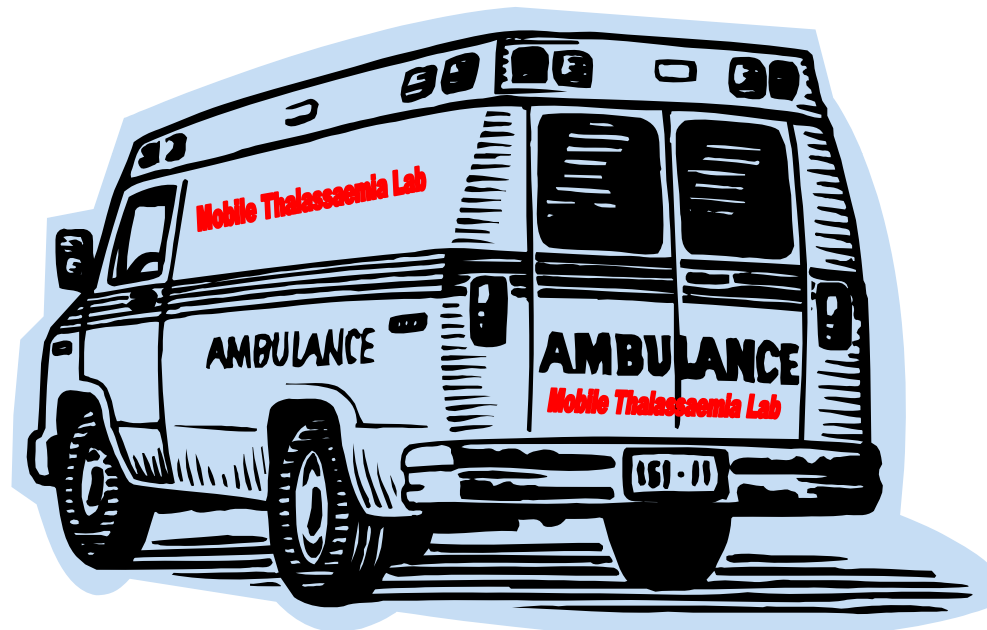
One Tube Osmotic Fragility Test

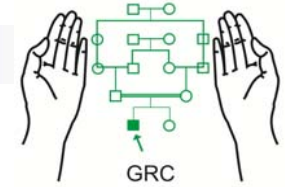
OTOFT		Total	Thal trait	Iron Def	Normal?
Positive (77%)	True	101	58	37	06
	False	33	-	10	23
	Total	134	58(43%)	47(35%)	29
Negative (22%)	True	36	-	06	30
	False	01	-	01	-
	Total	37	-	08	29
Borderline (1%)		02	-	01	01
Grand Total		173	58	56	59





Mobile Thalassaemia Service





Extended Family Screening

- It produces a high yield of carriers and at risk couples.
- Families often already understand the condition because of the affected child already present in the extended family.
- It avoids the problem of a low level of literacy, because information and personal experiences are communicated directly among families attending the centre with affected children.
- It avoids the problem of a weak health care infrastructure because it can be run from the centre where the disorder is diagnosed and treated.

