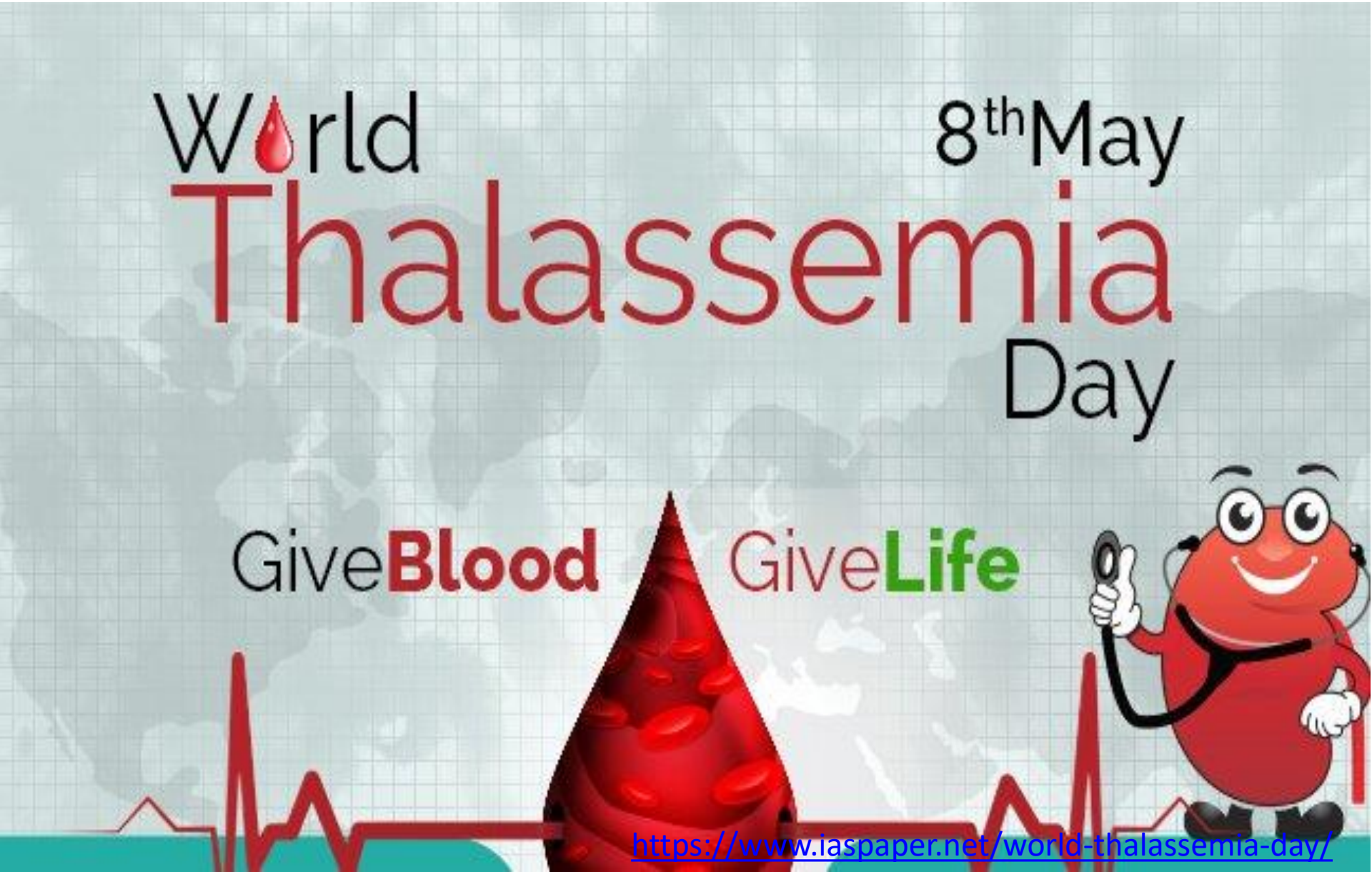
Overview of Thalassemia



https://www.iaspaper.net/world-thalassemia-day/

This year's (2020)theme is "Thalassemia Free India"

Chairperson
Clinical research committee
FOGSI

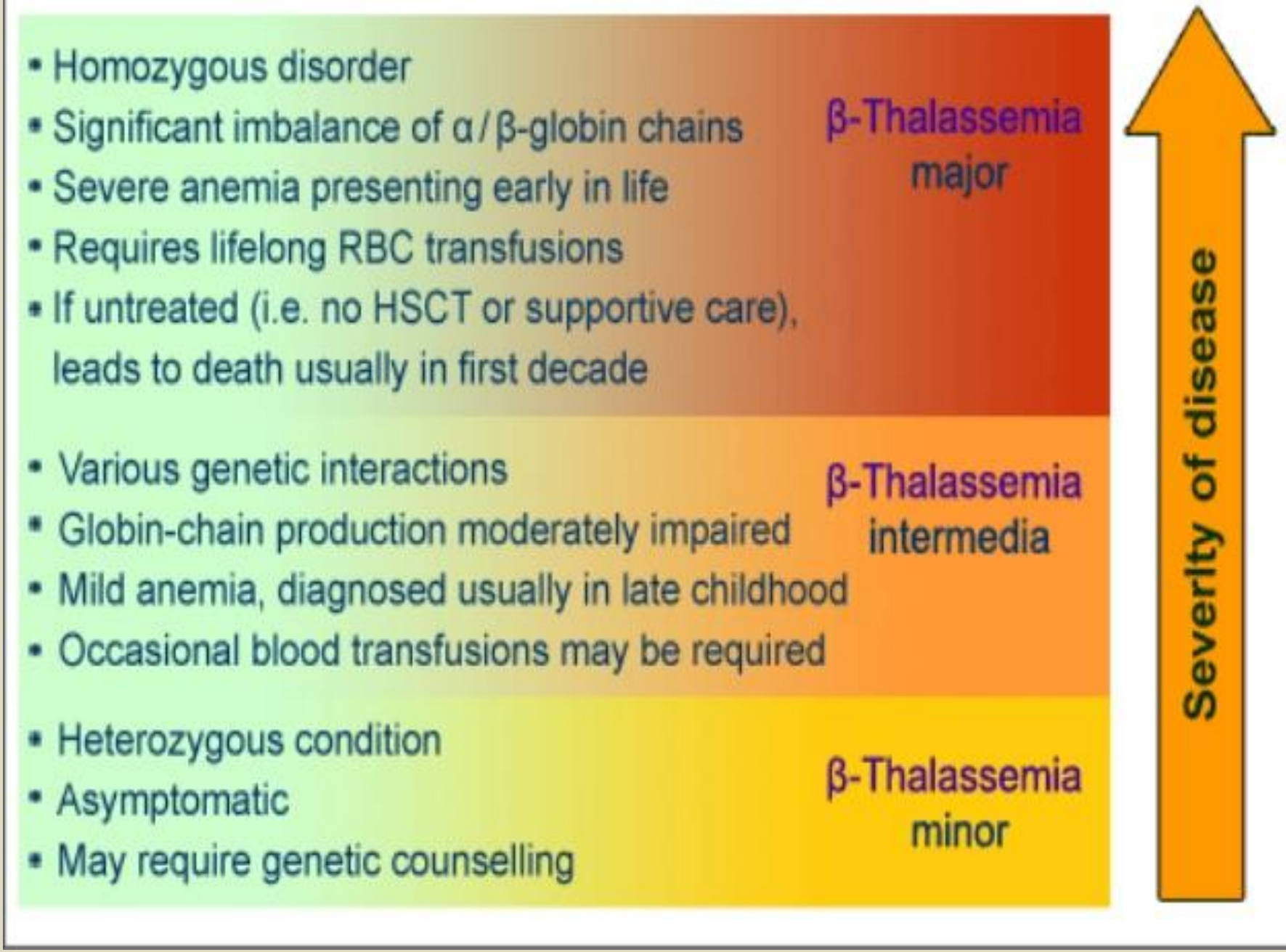
Dr.Sarita Anand
Senior GOGSian
Coordinator for Awareness on Thalassemia

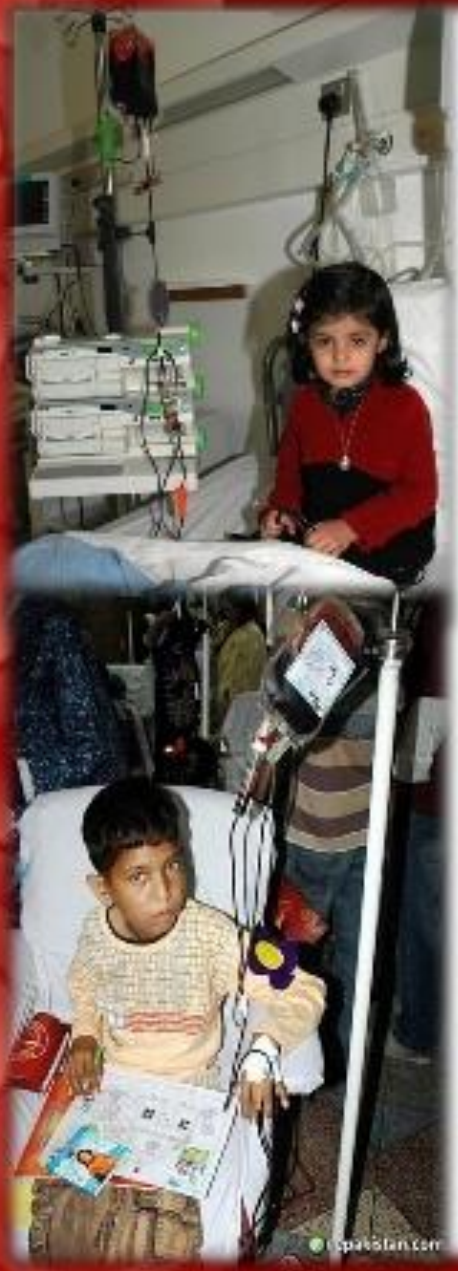
President
Ghaziabad Obstetric & Gynaecological Society

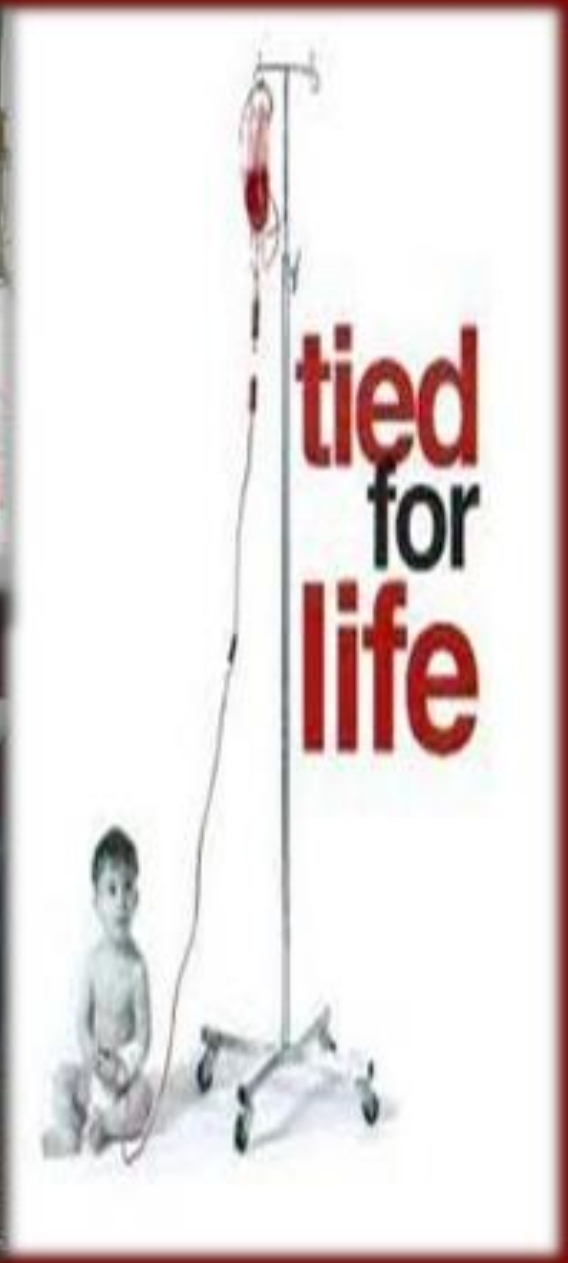
Have come together to bring you this awareness programme on Thalassemia

Thalassemia

- Derived from Greek word “thalasa”meaning ocean as patients were first identified along the coast of Mediterranean Sea
- By leading scientists Lee & Cooper in 1925.
- It is an inherited genetic blood disorder where patients cannot produce adequate healthy hemoglobin resulting in need for continuous blood transfusion.
- The Blood which saves their lives,creates an iron overload in the system.
- The excess iron must be chelated out of their system.
- The process is both painful and costly.


• Homozygous disorder
• Significant imbalance of α / β-globin chains
• Severe anemia presenting early in life
• Requires lifelong RBC transfusions
• If untreated (i.e. no HSCT or supportive care), leads to death usually in first decade
β-Thalassemia major
• Various genetic interactions
• Globin-chain production moderately impaired
• Mild anemia, diagnosed usually in late childhood
• Occasional blood transfusions may be required
β-Thalassemia intermedia
• Heterozygous condition
• Asymptomatic
• May require genetic counselling
β-Thalassemia minor
Severity of disease

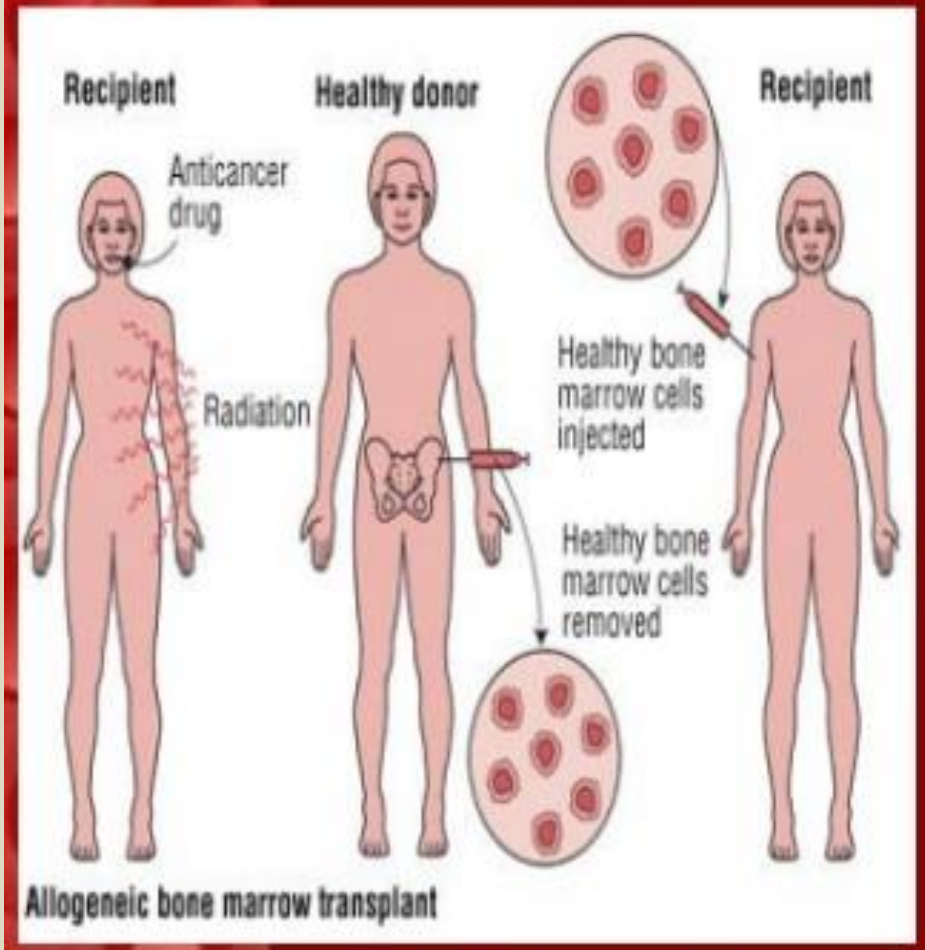


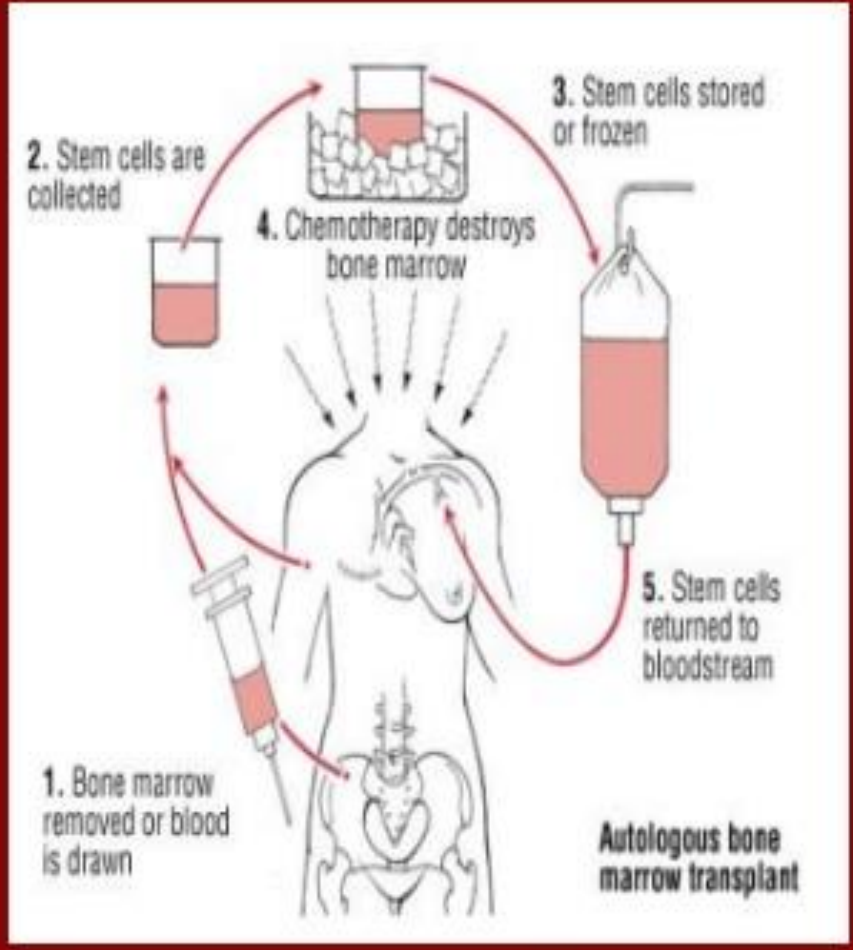


Thalassaemia child requires regular 3-4 weekly filtrated red blood cells transfusion.

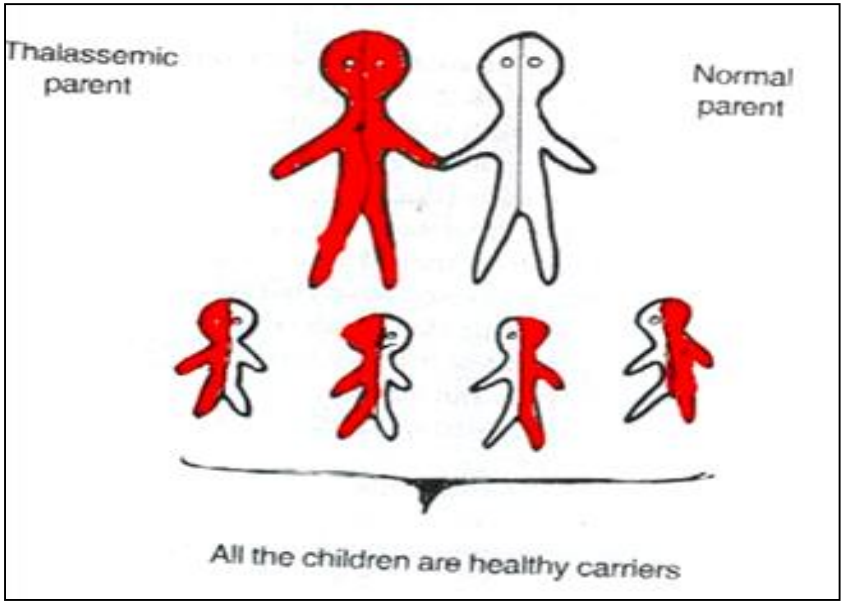
Iron chelation therapy

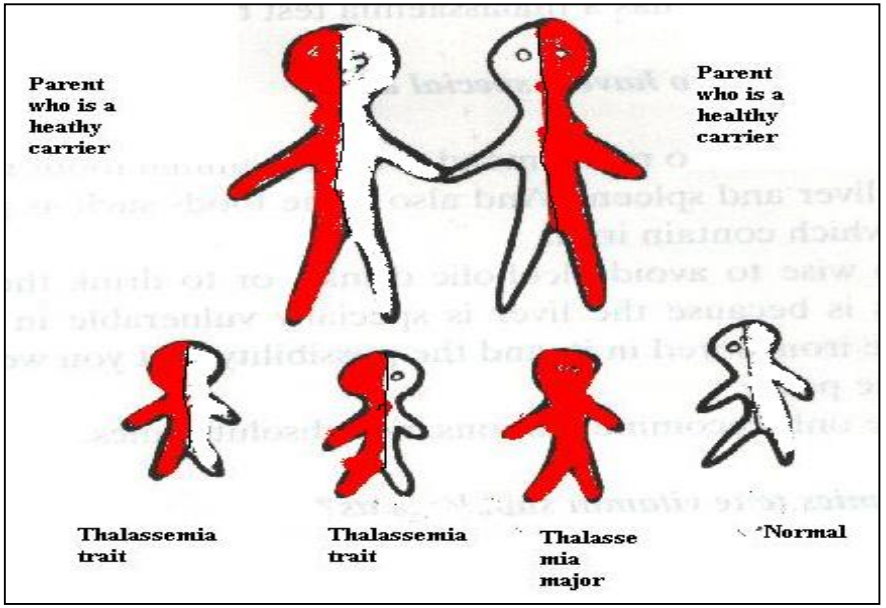
Repeated BT causes iron load.
Desferoxamine is Given S.C. for 8 hours every day for 5-6 days a week so as to chelate iron load.

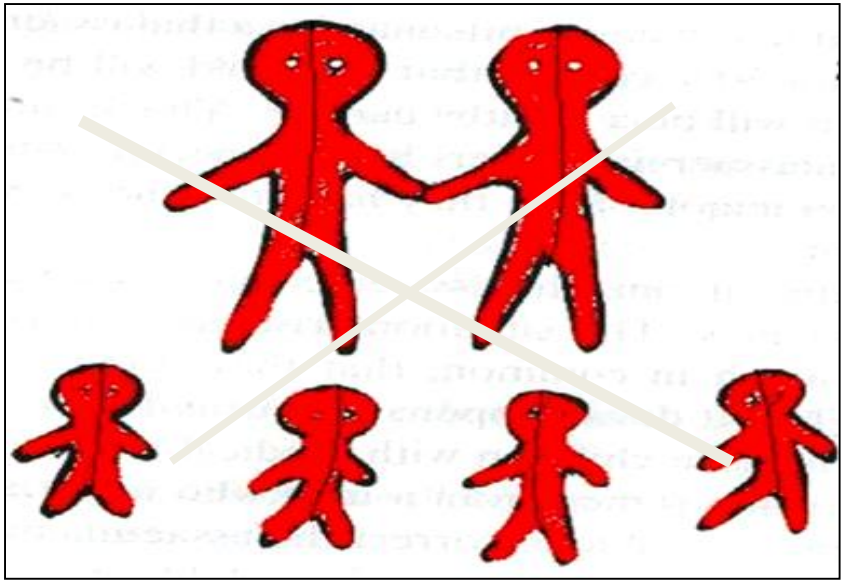


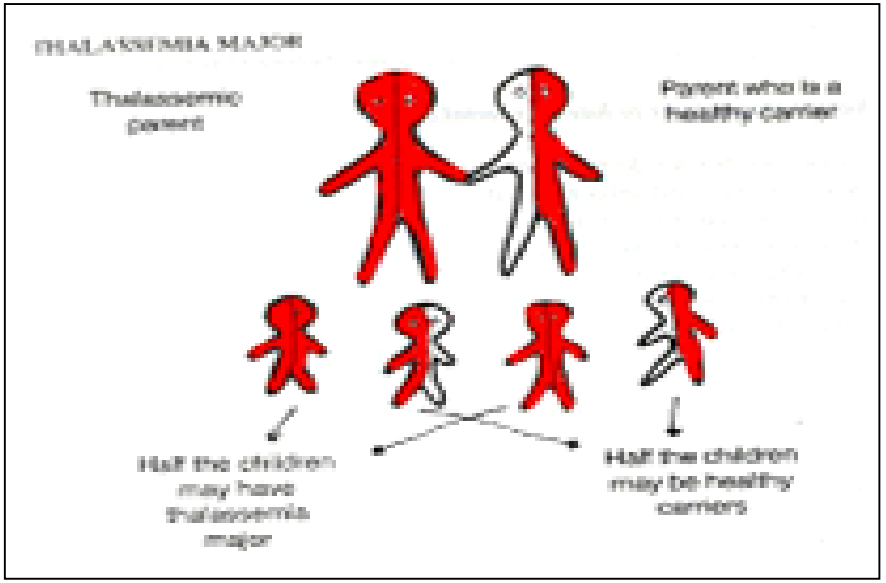


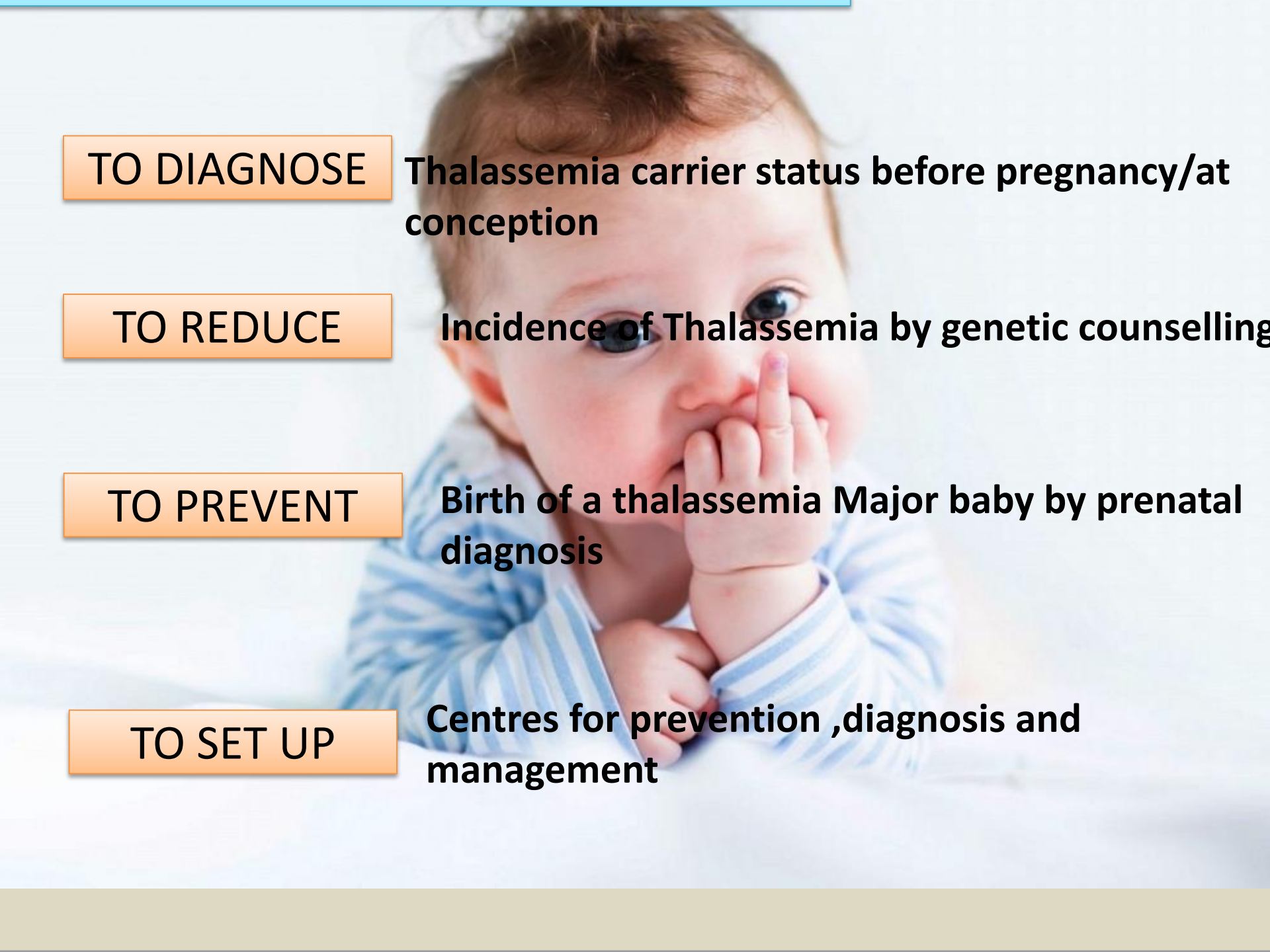
The only curative treatment is Bone marrow - stem cell Transplantation which cost around 10-15 lacs


Thalassemic parent
Normal parent
All the children are healthy carriers


Parent who is a heathy carrier
Parent who is a healthy carrier
Thalassemia trait
Thalassemia trait
Thalasse mia major
Normal




THALASSEMIA MAJOR
Thalassemic parent
Parent who is a healthy carrier
Half the children may have thalassemia major
Half the children may be healthy carriers



TO DIAGNOSE	**Thalassemia carrier status before pregnancy/at conception**
TO REDUCE	**Incidence of Thalassemia by genetic counselling**
TO PREVENT	**Birth of a thalassemia Major baby by prenatal diagnosis**
TO SET UP	**Centres for prevention ,diagnosis and management**

Baseline screening test

MCV < 75

MCH < 25

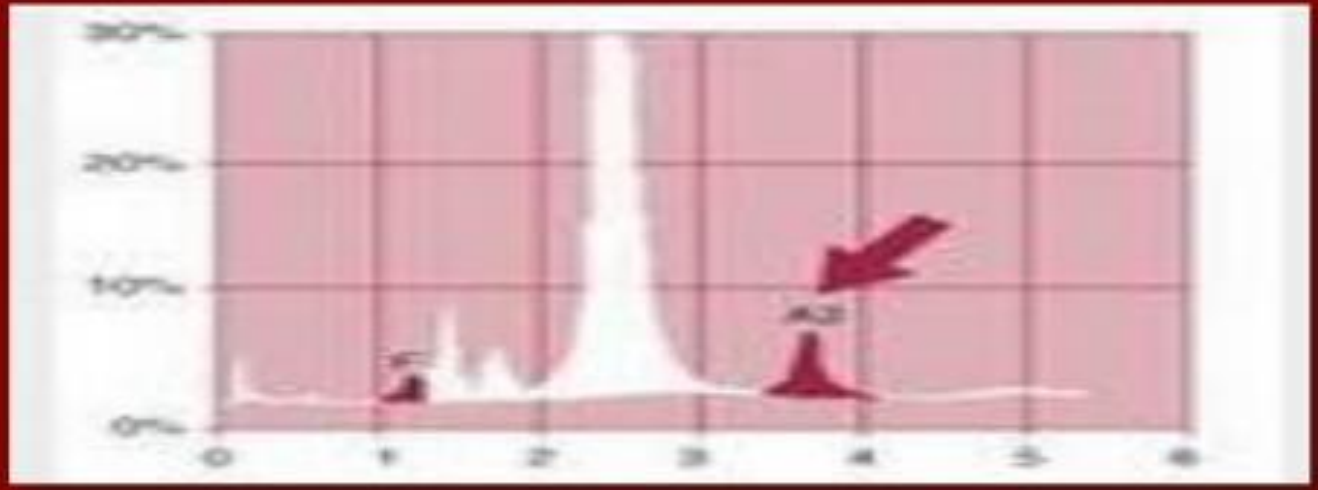
Mentzer Index $= \frac{MCV}{TRBC} < 13$

This Magic figure to be keep in mind for suspecting person is Thalassemia carrier

Second level screening test

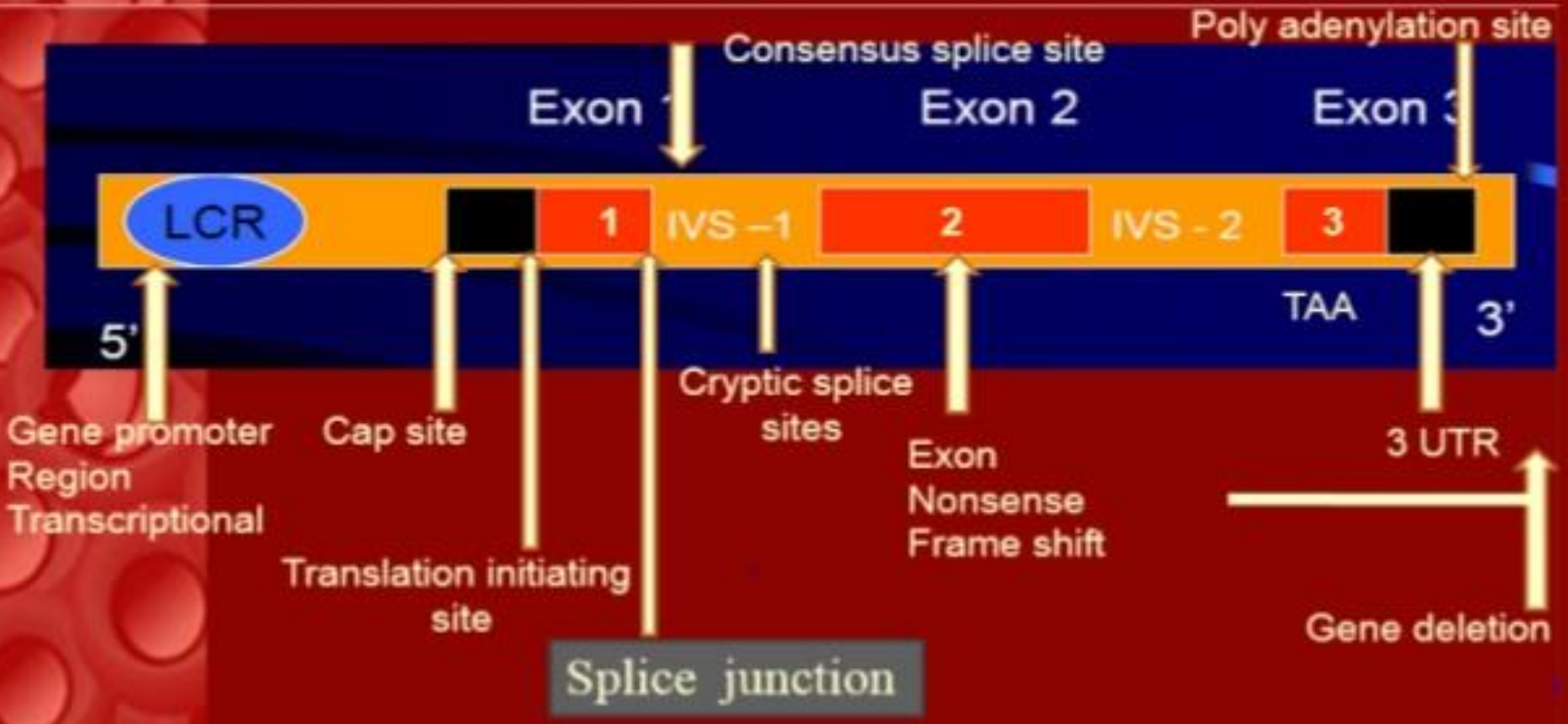
High performance liquid chromatography is the Gold standard test to estimate the HbA2 level for detecting carrier state

HbA2 > 3.5%
is suggestive of thalassemia carrier



Third level Confirmatory test

DNA analysis of the carrier for detection of thalassemia gene mutation is essential



CARRIER SHOULD NOT MARRY A CARRIER

MEDICAL KUNDLI MILAN

INSTEAD

OF

JANAM KUNDLI MILAN

As responsible citizens we should not be ignorant and should be able to take decision whether we wish to feed our child with milk or blood

NONINVASIVE

Maternal blood is collected for Isolation of Fetal DNA
fetal DNA is then tested for <u>Paternal mutant gene</u>

Paternal mutant gene Absent

Fetus is normal or carrier

Continue the pregnancy

Paternal mutant gene present

Fetus has 50% chance being Thalassaemia Major

Massively parallel DNA Sequencing done

Lets take pledge to screen ourselves for thalassemia

Lets take pledge to donate blood