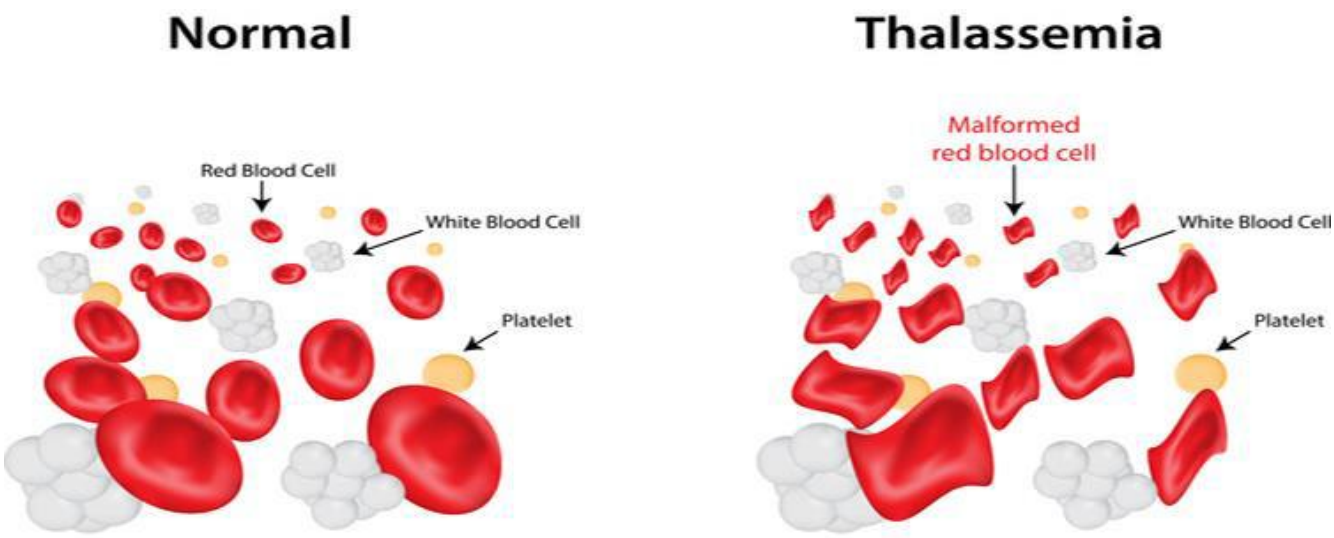
Alpha Thalassemia

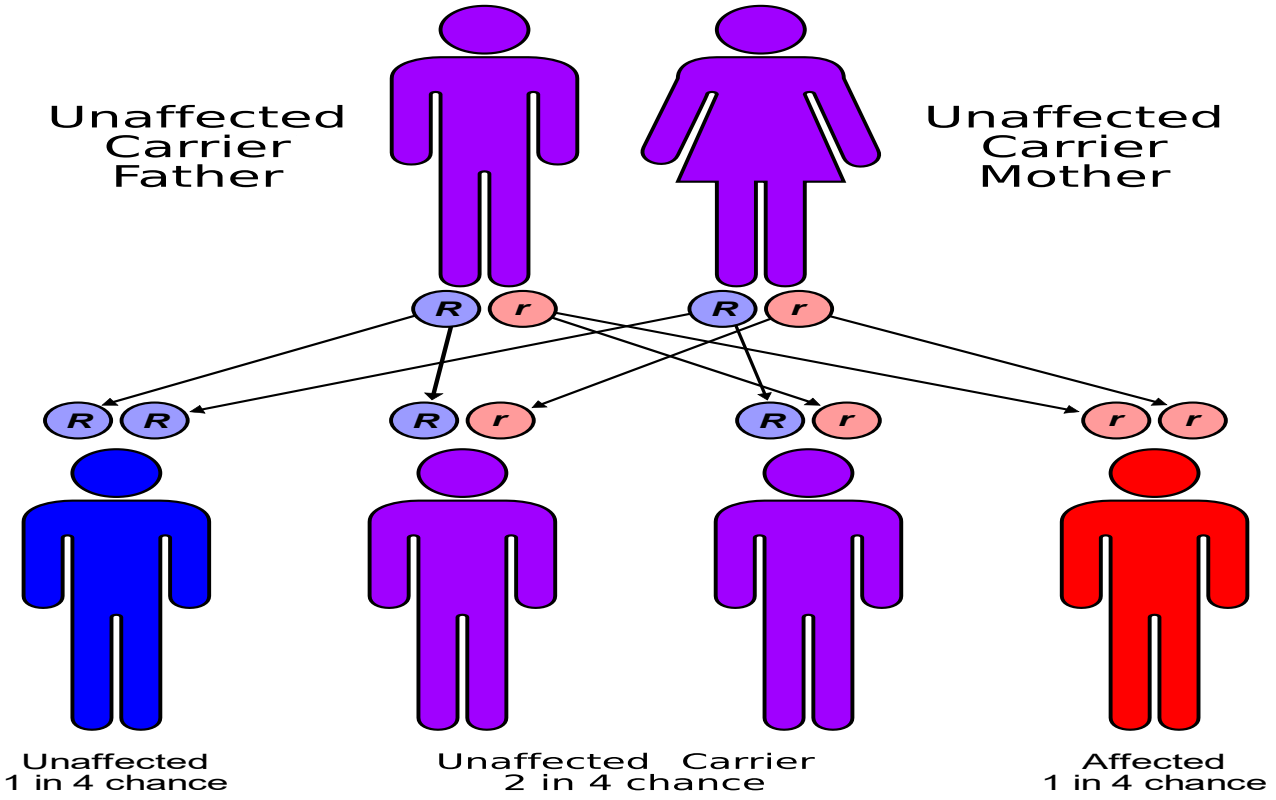
- Alpha thalassemia is a blood disorder that reduces the production of haemoglobin. Haemoglobin is the protein in red blood cells that carries oxygen to cells throughout the body.
- Two types of alpha thalassemia can cause health problems.
- In people with the characteristic features of alpha thalassemia, a reduction in the amount of haemoglobin prevents enough oxygen from reaching the body's tissues.
- Affected individuals also have a shortage of red blood cells (anaemia), which can cause pale skin, weakness, fatigue, and more serious complications.



Clinical Utilities:

ALPHA THALASSEMIA MUTATION DETECTION:

- Alpha Thalassemia PCR Chain is either due to small deletions (deletional mutations) or point mutations (non-deletional mutations) in the alpha globin gene.
- 95% of South Asian population show deletional mutations.
- The most common deletional mutations observed in South east Asian population are 3.7, 4.2, SEA, FIL and THAI.



Test Details:

Test Name	Test Code	MRP	TAT / Reported on
Alpha Thalassemia Mutation	SMO10120	4300	12th Working day by 7:00 p.m.
Alpha Thalassemia/ Alpha gene triplication copy number variations	SMO10351	11520	21st working day by 7:00 p.m.
Beta Thalassemia mutation	SMO10188	5500	5th Working day by 7:00 p.m.
Beta Thalassemia Mutation (13 mutations)	SMO10188	5500	5 days, if Sample received before 2:00 pm on Day 1
Metabolic screen TMS + 6 + Hb (CH, CAH, G6PD, CF, Galactosemia, Biotinidase, Sickle Cell Disease, Sickle Cell trait, Hb variants, B thalassemia)	RP10216	5890	8th Working Day by 7:00 p.m.
SES- Infectious Endophthalmitis PCR	SMO10160	32200	3rd working day if received before 1300hrs
Thalassemia Mutation (5 common mutations) Prenatal Specimen	SMO10418	17000	11th working days by 7:00 p.m.
Thalassemia Profile	RP10049	2200	3rd Working Day by 7:00 p.m.
Metabolic screen TMS + 6 + Hb (CH, CAH, G6PD, CF, Galactosemia, Biotinidase, Sickle Cell Disease, Sickle Cell trait, Hb variants, B thalassemia)	RP10216	5890	8th Working Day by 7:00 p.m.
Thalassemia Profile	RP10049	2200	3rd Working Day by 7:00 p.m.