

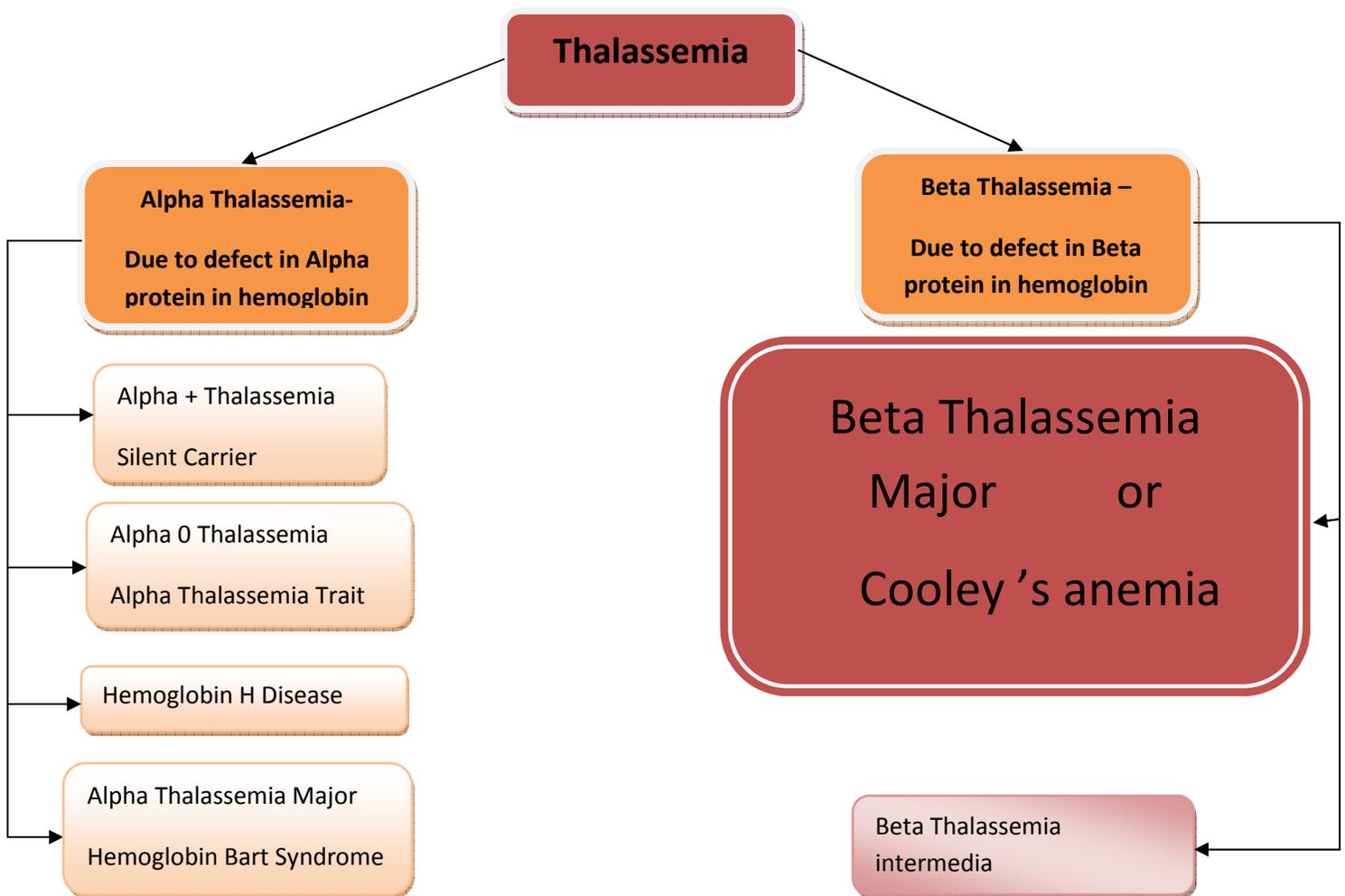
Everyone should know about Thalassemia

Thalassemia is a group of genetic blood disorder which is inherited through the generations of family which has either Thalassemia carrier or Thalassemia patient in the family.

Hemoglobin is a protein that is present in the Red Blood Cells (RBCs) of the blood. Hemoglobin carries oxygen in the blood which is important for functioning of different cells & tissues of body.

Hemoglobin has 2 different sets of proteins namely Alpha protein and Beta protein. Failure to produce either of these two or both the alpha & beta proteins results in formation of immature RBCs. Normal lifespan of RBCs is up to 120 days. In case of people having Thalassemia due to immature RBCs the lifespan of such RBCs is very less which results in early breakdown of such RBCs and the hemoglobin from such RBCs mixes up in the blood and gets deposited in liver, spleen and bone and gives rise to anemia which is called as Hemolytic anemia.

Thalassemia has different types depending upon the protein in which the genetic defects (mutations) are present. Different types of Thalassemia are shown below.



What are the Signs & Symptoms of Thalassemia?

As previously discussed, patients having Thalassemia has group of symptoms depending upon the type of Thalassemia and severity of anemia.

Alpha Thalassemia:

This type is more common amongst Indians and many people are silent carriers of Thalassemia which means they have genetic mutations in hemoglobin but without any symptoms.

Signs & symptoms of Alpha Thallasemia are as follows,

Type of Alpha Thalassemia	Symptoms
Alpha + Thalassemia (Silent carriers)	None
Alpha 0 Thalassemia	Mild Anemia
Hemoglobin H Disease	Symptoms usually develop in late childhood or early adulthood & this type can be treated with blood transfusion. <ul style="list-style-type: none">- Mild to moderate anemia- Pallor, weakness, fatigue- Enlargement of liver & spleen- Overgrowth of upper jaw & prominent forehead
Alpha Thalassemia Major	Usually babies with this type are still born or die shortly after birth and this condition is not treatable.

Signs & Symptoms of Beta Thalassemia:

Beta Thalassemia is also common amongst Indians.

Beta Thalassemia major manifests as mild to moderate anemia and often misdiagnosed as Iron deficiency anemia.

Beta Thalassemia Major or also known as Cooley's anemia is most severe form of Thalassemia which manifests as severe anemia, liver & spleen enlargements and other associated clinical problems and requires repeated blood transfusions.

How to diagnose Thallasemia?

If you have any family history of Thalassemia, there are chances that you might be the silent carrier of Thalassemia or might have actual Thalassemia.

You can come to know about family history of Thalassemia by asking following questions.

- 1) Does anybody from your family members are receiving repeated blood transfusion?
- 2) Does anybody from your family members has undergone bone marrow transplant treatment in past?
- 3) Is there a history of still birth of infant death in your family due to anemia?

If you get YES answer to any of the above questions you should consider yourself a potential candidate to undergo Thalassemia screening.

Thalassemia Screening

Every person who has signs & symptoms mentioned in above table or those who has family history should do a simple blood test called Complete Blood Count (CBC). It will cost nothing more than Rs.100 and can be done at any pathology lab even at rural places.

CBC measures all the components of blood indices and following table shows normal blood components with their corresponding normal value range in the blood.

Hemoglobin	12-17 gm% (males) 11-15 gm% (females)
White Blood Cells or Total Leucocytes counts (WBC or TLC)	5000-10000 per cubic mm.
RBCs count	4,50,000 - 6,00,000 (Male) per cubic mm. 4,50,000 - 5,50,000 (Female) per cubic mm. 5,00,000 - 7,00,000 (Newborn) per cubic mm.
Packed Cell Volume (PCV)	40 – 54% per 100 cc. (Male) 37 -47% per 100 cc (Female)
Mean Corpuscular Volume (MCV)	82-92 cubic microns
Mean Corpuscular Hemoglobin (MCH)	27-33 micrograms
Mean Corpuscular Hemoglobin Concentration	32-36%

MCV or mean corpuscular volume measures the average size of Red Blood Cells and MCH or Mean Corpuscular Hemoglobin Concentration measure amount of hemoglobin in each Red Blood Cell.

If both the MCV and MCH values of your blood count are less than the normal range, you are a potential candidate to undergo Thalassemia Screening Tests.

The next step in Thalassemia screening is to test for abnormalities of hemoglobin within the blood of the persons who has anemia and low MCH & MCV values. There are two types of hemoglobin Hb A and Hb F. Normal persons has 96-98% Hb A and 2-3% of Hb F.

Two types of tests are available for Hemoglobin analysis.

- 1) Hemoglobin electrophoresis
- 2) High Performance Liquid Chromatography.

Person should consult the physician first before undergoing such type of specialized testing.

Why it is important to know about the Thalassemia?

As we have seen earlier, Thalassemia is a genetically inherited blood disorder can be transmitted to the baby if any of the parents are either carrier or having Thalassemia.

Pre marital as well as pre conception counseling of all married couples is important because it can prevent the baby being born with deadly Thalassemia disorder.

Whom to consult?

Persons who has family history of Thalassemia or who are willing to undergo Thalassemia screening should consult the physician first. Newly married couples who intend have a child should consult gynecologist and undergo screening.

There are genetic counselors in metro cities who can also guide you about Thalassemia screening.

What are the treatment options for Thalassemia?

Patients having Thalassemia has different treatment options depending upon the type of Thalassemia they are having. These options includes,

- 1) Blood Transfusions
- 2) Bone Marrow Transplant
- 3) Family Planning and Chorionic Villous Sampling for Intrauterine diagnosis of Thalassemia

