North Delhi Pathology Clinic
27/5, Shakti Nagar, Nangia Park, Delhi-110007

# Thalassemia
## Major & Trait

**Dad !!**
**Have you**
**ever tested yourself**
**& Mom for Thalassemia**
**status to ensure that I am OK?**

Thalassemia is a type of anemia, which runs in families.

It can be of two types:

- Thalassemia Trait (Syn: Carrier, Minor)
- Thalassemia Major

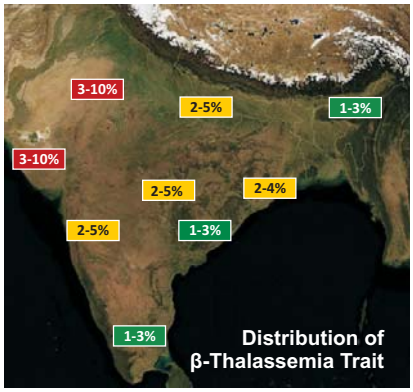
### Thalassemia Trait (Syn: Carrier, Minor)

Persons having Thalassemia trait usually have a very mild anemia. They lead normal lives and most of them do not even know that they carry it. The disease is only discovered when a person takes a blood test (hemoglobin chromatography). When two Thalassemia traits marry there are very high chances (25%) that they will bear a child who is Thalassemia major. In local population it is estimated that approximately 6 % (almost every 15th person) are having Thalassemia carriers is in Delhi.

### Thalassemia Major

A child with Thalassemia major appears healthy at birth but gradually becomes pale and develops an enlarged liver or spleen. These symptoms and signs generally occur within the first 3-12 months of life. Only treatment available as of now in India is multiple and frequent blood transfusions to maintain adequate hemoglobin level through out the life. All kinds of Thalassemia are heredity diseases and they are passed on in one of the three ways:

| Parents' Status | Possibility in Children |
|---|---|
| Both parents normal | All normal |
| One trait, other normal | 50% Thalassemia trait, 50% are normal, No Thalassemia major |
| Both parents thalassemia trait | 25% Thalassemia major, 50% Thalassemia trait, 25% normal |




### Available Blood tests for Thalassemia detection:

**Screening test:**

Hemogram on automated hematology analyzer (Hemoglobin, Total RBC Count specially MCV & RDW-SD and other absolute values) | Iron Studies | S. Ferritin.

All suspicious cases must undergo confirmatory test.

**Confirmatory test:**

Hemoglobin Chromatography by HPLC methodology.

**Further tests:**

Parental & Genetic Studies

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# Thalassemia & other Hemoglobinopathies:

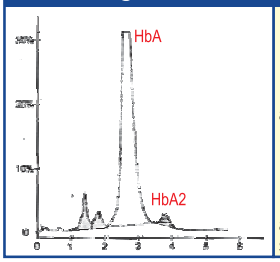
## Common HPLC Patterns in North Indian Population

Thalassemia and hemoglobinopathies are autosomal recessive conditions affecting the quantity and quality, respectively, of hemoglobin molecules within red blood cells.

Since they are genetic conditions, it is important to prevent these conditions by detecting them early, by performing a simple HPLC test on blood early, preferably before marriage. Cation exchange HPLC has emerged as the method of choice for testing. It is possible to quantify Hb A2, Hb F, Hb A along with screening hemoglobin variants like Hb S, Hb D, Hb E and Hb C in a single, highly reproducible system, making it an excellent technology to screen for hemoglobin variants and hemoglobinopathies along with the thalassemias. Various studies have shown **equivalence or superiority over electrophoretic** methods.

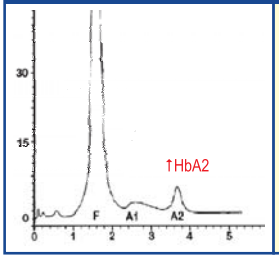
Common Hemoglobinopathies prevalent in North India and their Chromatographic patterns are as follows:

### Chromatogram of Normal Subject



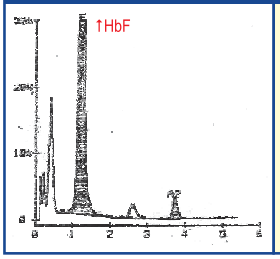

**Phenotype:** Normal.
**Ethnicity:** Seen in all normal adults worldwide.
**Lab Findings:** There is no anemia or reticulocytosis. MCV, MCH are usually normal.
**HPLC Pattern**: HbA2 is less than 3.5%. No abnormal/ unknown peak seen.

### Beta thalassemia trait



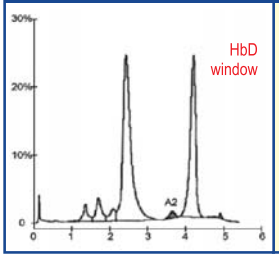

**Phenotype:** Normal or mildly anemic.
**Ethnicity:** Prevalent in North & Western India (3-7%).
**Lab Findings:** Microcytic hypochromic RBCs, MCV < 75 fl & MCH reduced < 25pg. RDW normal RBC count increased.
**HPLC Pattern:** Elevated HbA2 > 4%.

### Beta thalassemia major



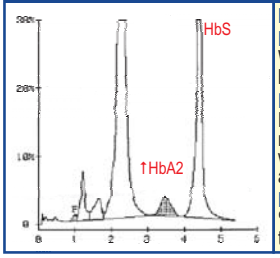

**Phenotype:** Severe anemia at 3-6 months.
**Lab Findings:** Presence of Severe hypochromia, microcytosis, poikilocytosi, polychromasia, target cells and nucleated RBCs.
**HPLC Pattern**: Elevated HbF >85%. Reduced HbA <3%.

### Hb D Punjab Heterozygous



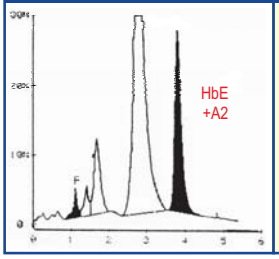

**Phenotype:** Normal.
**Lab Findings:** Normal Hb & RBC indices.
**HPLC Pattern:** HbD window 30-45%. HbF normal, HbA2 Lower side 0.9-2.5%.

### Hb S Heterozygous



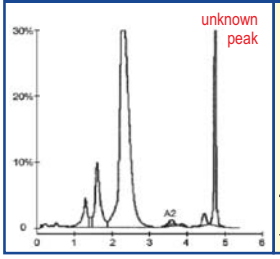

**Phenotype:** Asymptomatic.
**Ethnicity:** Prevalent in Western, central and eastern belts of India.
**Lab Findings:** Normal HB. Hypochromia, microcytosis or macrocytosis if association with thalassemia or nutritional anemia.
**HPLC Pattern:** HbS: 30 & 40 %. HbA2 may be elevated due to co-elution of Glycated HbS.

### Hb E Heterozygous




**Ethnicity:** Prevalent in Northeastern India.
**Lab Findings:** No anemia or reticulocytosis is noted. MCV and MCH may be low.
**HPLC Pattern:** HbE is about 30% and elutes in HbA2 window. HbF: normal.

### Hb Q India Heterozygous




**Phenotype**: Normal.
**Lab Findings:** Normal. MCV & MCH may be marginally reduced.
**HPLC Pattern:** Abnormal hemoglobin is seen as unknown peak in the retention time of window of 4.77 min + 0.01 constituting usually 8.7- 23.3% of the total hemoglobin.

There are other rare hemo-globinopathies prevalent in north India like Hb Lepore, HbD Iran, various forms of Hb J, HbD and other even rarer double heterozygous forms. Majority of these conditions are also easily detected using HPLC.

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