



Thalasseмииs

Thalasseмииs are hereditary disorders characterized by reduction in the synthesis of the alpha and beta variety of globin chains. Reduced globin chain synthesis causes reduced Hemoglobin synthesis that eventually produces a hypochromic microcytic anemia because of defective haemoglobinisation of red blood cells.

HOW TO DIAGNOSE?

1. Microcytosis out of proportion to the degree of anemia. (Hb 11.5 Gm % & MCV – 65fl)
2. Positive family history
3. Abnormal RBC morphology with target cells, microcytes and acanthocytes
4. In beta thalasseमीs elevated levels hemoglobin of A₂ & F (Hb electrophoresis)

PATHOPHYSIOLOGY: Normal adult Hb is primarily Hb A which represents about 98% of circulating Hb. The remaining component comprises of HbA₂ & Hb F (Fetal Hb).

THALASSEMIAS ARE OF 2 TYPES:

1. Alpha thalasseमीs – seen primarily in South-East Asia and China. These patients may be either essentially normal and with normal life expectancy and performance status and may have a very mild microcytic anemia. The other end of spectrum is a hemoglobin H disease with haemolytic anaemia of variable severity. Patient may look pale and may

even present with splenomegaly. The difference in presentation status is to do with alpha globin genes are present. A detailed discussion is out of purview of this article.

2. Beta Thalasseमीs – mainly affects Europeans and Greek and to a lesser degree Chinese and other Asians. Amongst Indians, Marwari, Kutchi, Parsi and Sindhi communities. Patients homozygous for thalasseमीs have the syndrome of Thalasseमीs Major. Affected children are normal at birth but within first year of life develop severe anemia requiring transfusion. Signs of Thalasseमीs generally manifest after 6 months of age as this is the time Hb synthesis switches from Hb F to Hb A. Clinical problems that a child may present with are, growth failure, bony deformities, hepatosplenomegaly and jaundice. The clinical course can be favourably altered with repeated blood transfusions but the overload of iron from transfusion (hemosiderosis) may lead to heart failure, cirrhosis and major endocrinopathies.

Patients heterozygous for beta-Thalasseमीs have Thal Minor. These patients have a mild microcytic anemia which is not clinically significant. Both parents having Thal minor stand a high chance of one of the children getting Thal Major.

	Hb A	Hb A ₂	Hb F
Normal adults	97-99%	1-3%	Less than 1%
Thal Minor	80-95%	4-8%	1-5%
Thal Major	0-10%	4-10%	90-96%
Thal Intermediaris	0-30%	0-10%	6-100%



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Friend: What are you looking at?

Banta Singh: I know your Password, hee, hee.

Friend: All right, what is my Password if you saw it?

Banta Singh: four asterisks!

