



What is Thalassemia?

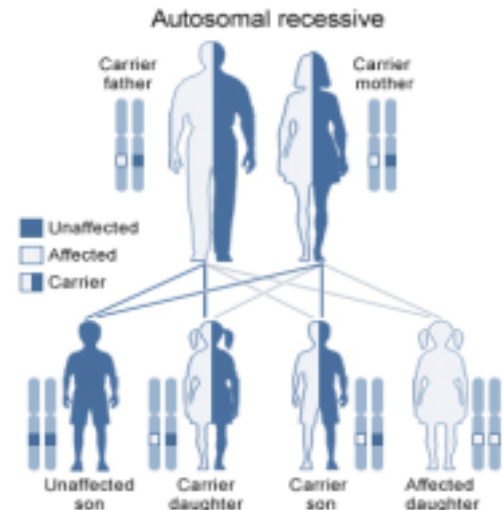
Thalassemia is the name of a group of genetic blood disorders characterized by anemia due to enhanced red blood cell destruction. Hemoglobin, the oxygen-carrying component of the red blood cells consists of two different proteins, an **alpha** and a **beta**. If the body doesn't produce enough of either of these two proteins, the red blood cells become defective and cannot carry sufficient oxygen. The resulting anemia is usually severe with several health problems like enlarged spleen, bone deformities, fatigue and requires regular life-long transfusion, therapy and medical supervision

How is Thalassemia inherited?

Genes are the hereditary units that come in pairs. They are passed on from parents (one from each) to children from one generation to the next.

Thalassemia belongs to the category of genetic disorders which are expressed when both genes in the pair are affected. Such inheritance is called autosomal Recessive inheritance. Genes responsible for haemoglobin synthesis are defective (mutated) or are missing altogether in the disease. Hundreds of mutations have been reported on globin genes causing Thalassemia.

Most often, the parents of a child with Thalassemia are not affected but are carriers with mutation in only one copy of the gene. Carriers have 25% chance of conceiving a child with Thalassemia.



How common is Thalassemia in India?

Thalassemia is a common inherited disease in the world. India accounts for 10% of the total world thalassemia population and approximately 1 in 30 in the general population is carrier of the mutated gene. Every year about 15,000 infants are born with haemoglobinopathies in India. Nearly 28 mutations are reported in beta Thalassemia Indian population of which eight accounts for 95% of the cases. Alpha Thalassemia is generally caused by deletions on alpha globin gene. Mutations are specific to population and state specific mutations are reported.

What is Thalassemia Gene test?

The Thalassemia genetic test or DNA analysis detects the mutation / deletion / defect in alpha or beta globin genes that causes Thalassemia. A small blood sample is needed. The lab uses sophisticated technology and high end equipment to identify and interpret the test results. Novel, rapid and sensitive methods are available to diagnose the condition both in the affected individual and carrier. For the affected child the test is performed just before the blood transfusion or after 3 weeks of blood transfusion.

Necessity of Gene testing for Thalassemia

Routine screening tests like CBP, blood smear, haemoglobin electrophoresis, and iron studies can identify Thalassemia in most of the cases but absolute confirmation is done by genetic test which identifies the existence of the root cause of disease. In cases where diagnosis between iron deficiency and Thalassemia carrier is doubtful 'genetic testing' can ascertain accurate diagnosis. Also, genetic tests help in identifying the type and severity of Thalassemia. Most importantly, for precise carrier testing and diagnosis of the condition in the fetus genetic testing is absolutely must.

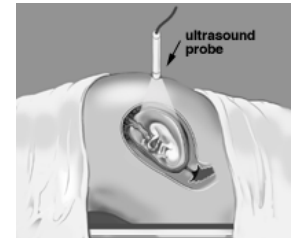
For this reason, affected family member and carriers should get genetic testing done in a lab that offers maximum sensitivity for the test. It also helps in providing appropriate genetic counseling to patients.

Sensitivity of Gene testing

The more number of mutations tested the better is the sensitivity. For example if the lab is testing for 2 mutations they may miss 25% of the gene defects. If the lab tests 4 mutations the chances of missing the defects can come down to 15%. GeneTech tests for common 8 Indian mutations for beta thalassemia and major deletions in alpha thalassemia offering sensitivity of more than 95%. The lab also offers specialized tests for rare defects. The sensitivity of the test also depends on the quality procedures and expertise available in the lab.

How accurately can Thalassemia be diagnosed in the fetus?

The test is extremely sensitive and accurate if the mutation is already identified in the affected family member or in parents (carriers). Prenatal genetic tests are widely in use to diagnose the condition in the fetus before birth worldwide. It is done by analyzing fetal DNA by Chorionic Villus sampling at 11 weeks of gestation or by amniocentesis at 16th week of gestation. It is important to rely on sophisticated lab with focus and genetic expertise. GeneTech has the best prenatal diagnostic facilities in the country today.



What is the treatment available for Thalassemia?

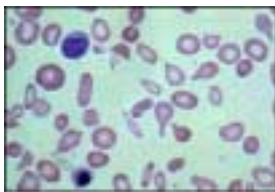
Specific treatment for alpha thalassemia will be determined by the doctor based on age, overall health, medical history, extent of the disease, tolerance for specific medications, procedures, or therapies. Treatment may include daily doses of folic acid, blood transfusions (as needed), medication to decrease amount of iron in the body, surgical removal of the spleen and bone marrow / stem cell transplant in severe forms.

What is Gene Therapy?

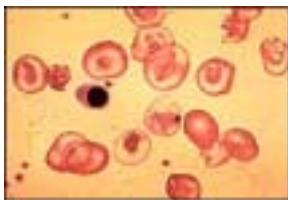
Gene Therapy involves creation of a mechanism for delivering corrected genes into cells. Specially-engineered viruses (known as retroviruses) are used to transport the corrected genes to cells. Multiple studies are under way to address various issues of gene therapy like tissue or cell targeting, other effects etc. Replacing the defective genes holds the greatest promise for a lifetime cure for Thalassemia in future.

How can Genetic Counseling help?

Genetic counseling helps families to understand the genetic aspects of the disorder. The process helps in identifying families at risk, investigate the problem present in the family, interpret information about the disorder, analyze inheritance patterns and risks of recurrence, and review available options like prenatal diagnosis. Genetic counselors provide supportive counseling to families and help parents understand and reach decisions about what to do next



Blood picture of
alpha Thalassemia



Blood picture of
beta Thalassemia



Blood picture of
Sickle cell Anemia

What are the different types of Thalassemia?

Alpha Thalassemia:

People whose hemoglobin does not produce enough alpha protein have alpha thalassemia. The common types are *Silent carrier* state (apparently normal), *Alpha Thalassemia Trait* or Mild Alpha Thalassemia (mild anemia that is often mistaken with iron deficiency), *Hemoglobin H Disease* (Severe form of anemia and health problems).

Hydrops Fetalis or *Alpha Thalassemia Major* is the condition where the abnormal hemoglobin called hemoglobin Barts is produced. Most individuals with this condition die before or shortly after birth.

Beta Thalassemia:

People whose hemoglobin does not produce enough beta protein have beta thalassemia. There are three types are *Thalassemia Minor* or Thalassemia Trait or Thalassemia Carrier (healthy with possible mild anemia), *Thalassemia Intermedia* (moderate to severe anemia including bone deformities and enlargement of the spleen) and *Thalassemia Major* or Cooley's Anemia (most severe form, life-threatening anemia, requiring regular blood transfusions and extensive ongoing medical care).

Haemoglobin E Beta Thalassemia:

Hemoglobin E is common abnormal hemoglobin and individuals present a moderately severe anemia which is similar in symptoms to beta thalassemia intermedia.

Sickle Beta Thalassemia:

This condition is caused by a combination of beta thalassemia and hemoglobin S and results in red blood cells that are defective sickle shaped. The condition varies from moderate to severe type of anemia.

Please contact our Genetic Counselors for more information on Thalassemia

