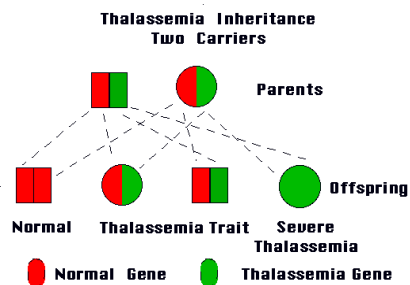


Beta thalassemia is a common blood disorder, affecting thousands of infants each year worldwide. It is caused by a reduced amount of the normal adult form of hemoglobin when beta-chains are non-functional or are present in reduced amounts. It can range in severity from severe anemia and hepato-splenomegaly in the first two years of life requiring blood transfusions to a much milder anemia presenting later in life. 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.

Inheritance

β Thalassemia is inherited in an autosomal recessive pattern; two copies of the gene must be mutated for a person to be affected by the disorder. Most often, the parents of a child with thalassemia are not affected but are carriers of one copy of the altered gene. Carriers have 25% chance of conceiving a child with thalassemia.



Molecular Pathology

Mutations in the HBB gene can reduce or abolish the production of beta-hemoglobin. More than 200 different mutations, most of which are single base changes in and around the β globin gene are known to cause β thalassemia.

Common Indian mutations (Specific to different states in India)

Nearly 28 mutations in beta globin gene have so far been recorded in India among which eight account for 95% of the cases (IVS1-5 (G→C), IVS1-1 (G→T), CD 8/9 (+G), CD 41/42 (-CTTT), CD 15, HbE, HbS and del 619bp). GeneTech's Thalassemia testing panel allows detection of common mutations specific to different state population in India.

Technique

Amplification Refractory Mutation System (ARMS) is a novel, rapid and sensitive tool used to detect not only mutations in affected individuals but also for carrier testing and prenatal diagnosis of β Thalassemia.

Prenatal diagnosis

With no effective treatment available and huge financial burden on families, prenatal diagnosis by CVS (10th week of gestation) or amniocentesis (16th week of gestation) are the most appropriate preventive measures to contain the disease and reduce the load of mutant alleles in gene pool.

Genetic counseling

Carriers of Thalassemia gene mutations will benefit from genetic counseling. It helps them understand the risk, complex possibilities, future therapy, recurrence risk and available prenatal options for prevention.

GeneTech's Thalassemia Test Panel

GeneTech offers Thalassemia testing for 8 common mutations on beta globin gene and two major deletions of alpha chain. 4ml of blood from each partner or affected individual would be required for testing. Prenatal samples include CVS, amniotic fluid or cord blood. The lab offers inhouse prenatal sampling facility. However, if sampling is done elsewhere, please coordinate the collection and transportation with GeneTech laboratory.

Sample report showing IVS1-5 heterozygote condition

