

THALASSEMIA BETA, MUTATION ANALYSIS

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MUTATION ANALYSIS	
TYPE OF MUTATION	RESULT (Homozygous Mutation Detected/ Heterozygous Mutation Detected/ Mutation Not Detected)
cap+1 [A>C]	
Codon 5 [-CT]	
Codon 6 [A>T] HbS	
Codon 8 [-AA]	
Codon 8/9 [+G]	
Codon 15 [TGG>TAG]	
Codon 16 [-C]	
Codon 22 [7bp del]	
Codon 30 [G>C]	
IVS 1.1 [G>A]	
IVS 1.1 [G>T]	
IVS 1.5 [G>C]	
IVS 1.6 [T>C]	
IVS 1.110 [G>A]	
IVS 1-25 [25bp del]	
Codon 36/37 [-T]	
Codon 39 [C>T]	
Codon 41/42 [-TTCT]	
Codon 44 [-C]	
IVS 2.1 [G>A]	
IVS 2.745 [C>G]	
619bp del	

Interpretation

RESULT	REMARKS
Homozygous mutation detected	Both copies of the gene carry mutation
Heterozygous mutation detected	One copy of the gene carries mutation
Mutation Not Detected	Both copies of the gene carry the wild type trait

Note

1. This test detects 22 most common mutations found in the Indian and Middle east region. It does not detect all the mutations in Beta Globin gene.
2. Test conducted on Whole blood for Postnatal Mutation analysis or Amniotic Fluid for Prenatal Mutation Analysis
3. Genetic Counseling available with prior appointment at National Reference Laboratory, New Delhi

Comments

Beta (β) thalassemia is an autosomal recessive disorder due to mutations in the HBB gene on chromosome 11. Severity of the disease depends on the nature of the mutation which is as follows:

β - Alleles without a mutation that reduces formation of β chains

β^0 - Mutations that prevent any formation of β chains

β^+ - Mutations that allow some formation of β chains

In all these cases there is a relative excess of α chains, but these do not form tetramers: rather, they bind to the red blood cell membranes, producing membrane damage, and at high concentrations they form toxic aggregates.

Depending on the Homozygous or Heterozygous state, Beta Thalassemia can be classified as:

Classification	Remarks	Alleles
Thalassemia minor	Only one β globin gene bears a mutation	β^+/β or β^0/β
Thalassemia intermedia	Condition intermediate between the major and minor forms.	β^+/β^+ or β^0/β^+
Thalassemia major	Both β globin genes bear a mutation.	β^0/β^0