Mutation Not Detected

Interpretation

<table>
<thead>
<tr>
<th>RESULT</th>
<th>REMARKS</th>
</tr>
</thead>
<tbody>
<tr>
<td>Homozygous mutation detected</td>
<td>Both copies of the gene carry mutation</td>
</tr>
<tr>
<td>Heterozygous mutation detected</td>
<td>One copy of the gene carries mutation</td>
</tr>
<tr>
<td>Mutation Not Detected</td>
<td>Both copies of the gene carry the wild type trait</td>
</tr>
</tbody>
</table>

Note

1. This assay detects more than 100 different mutations in the Promoter region, Exon 1, IVS-I & Exon 2 and part of IVS-II. It also detects the deletion of 619 bp in IVS II and Exon 3. This assay does not detect variants in other parts of this gene.
2. Presence of PCR inhibitors in the sample may prevent DNA amplification.
3. This is an in-house developed assay.
4. Test conducted on Whole blood for Postnatal Mutation analysis and Amniotic Fluid for Prenatal Mutation Analysis
5. 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
β0- 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 and Remarks

<table>
<thead>
<tr>
<th>Classification</th>
<th>Remarks</th>
<th>Alleles</th>
</tr>
</thead>
<tbody>
<tr>
<td>Thalassemia minor</td>
<td>Only one Beta globin gene bears a mutation</td>
<td>Beta+/Beta or Beta0/Beta</td>
</tr>
<tr>
<td>Thalassemia intermedia</td>
<td>Condition intermediate between the major and minor forms.</td>
<td>Beta+/Beta+ or Beta0/Beta+</td>
</tr>
<tr>
<td>Thalassemia major</td>
<td>Both Beta globin genes bear a mutation</td>
<td>Beta 0/Beta 0</td>
</tr>
</tbody>
</table>

The distribution of beta thalassemia gene is not uniform in the Indian subcontinent. The highest frequency of beta thalassemia trait is reported in Gujarat, followed by Sindh, Punjab, Tamil Nadu, South India and Maharashtra. Beta Thalassemia is common among Sindhi, Gujarati, Parsee, Punjabi Hindus, Lohanas and Teli communities of Indian subcontinent.

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**Important Instructions**

- Test results released pertain to the specimen submitted.
- All test results are dependent on the quality of the sample received by the laboratory.
- Laboratory investigations are only a tool to facilitate in arriving at a diagnosis and should be clinically correlated by the Referring Physician.
- Sample repeats are accepted on request of Referring Physician within 7 days post reporting.
- Report delivery may be delayed due to unforeseen circumstances. Inconvenience is regretted.
- Certain tests may require further testing at additional cost for derivation of exact value. Kindly submit request within 72 hours post reporting.
- Test results may show interlaboratory variations.
- The Courts/Forum at Delhi shall have exclusive jurisdiction in all disputes/claims concerning the test(s) & or results of test(s).
- Test results are not valid for medico legal purposes.
- Contact customer care Tel No. +91-11-39885050 for all queries related to test results.
- Sample drawn from outside source.

# Not in NABL scope