| | | | |
|---|---|---|---|
| Patient Name | **: Dummy** | | |
| DOB/Age/Gender | : 23 Y/Female | Sample Collected | : Mar 19, 2024, 12:24 PM. |
| Patient ID / UHID | : XXX | Report Date | : Apr 10, 2024,11:22 AM. |
| Referred By | : Dr. XXX | Barcode No | : XXX |
| Sample Type | : Whole blood EDTA | Report Status | : Final Report |

## Alpha Thalassemia Mutation Screening (3 Common Mutation)

**RESULTS :**

**THALASSEMIA ALPHA MUTATION ANALYSIS :** Mutation Not Detected

**Kindly note:**- Genetic counselling and Clinical correlation -CBC peripheral blood picture,history of blood transfusion, family history of similar complaints,and other laboratory/therapeutic details - recommended..

**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. The diagnostic sensitivity of this test is estimated to be close to 90%, since ~90% of all alpha-thalassemias are caused by deletions such as -α3.7, -αα 4.2, --SEA, --MED1, --MED2, (-α)20.5,--FIL, --THAI, --DUTCH1 etc but might not be able to distinguish between some of them. In addition, this test detects the Hb Constant spring mutation and the deletion in HS40 regulatory element.Approximately 10% of the genetic defects in the alpha-globin gene cluster result from small (point) mutations, most of which, except for the Hb Constant Spring mutation, will not be detected
2. Presence of PCR inhibitors in the sample may prevent DNA amplification
3. Analytical performance can be compromised by SNPs or other polymorphisms (e.g. indels) in the DNA target sequence
4. Test conducted on Whole blood for Postnatal Mutation analysis.
5. Genetic Counseling available with prior appointment at National Reference Laboratory,

**Comment:**
Normally there are 4 copies of alpha globin genes, two copies on each chromatid of chromosome 16. Alpha globin chains are essential in the synthesis of both fetal and adult hemoglobin. Alpha thalassemia syndromes results from deletion of large alpha globin gene segment from unequal cross-over or recombination and less frequently from mutations. Both alpha globin gene deletion haplotypes (-/+) and (-/-) occur equally in southeast Asians whereas (-/-) haplotypes is much less common in Mediterraneans and rare in Africans.

**Interpretation :**

| AlIELES AFFECTED | DESCRIPTION | GENOTYPE |
|---|---|---|
| One | This is known as alpha thalassemia silent and with this type, the effect on hemoglobin synthesis is minimal. Three α-globin genes are enough to permit normal hemoglobin production, and no clinical symptoms present. It occurs due to a deletion or non-deletion mutation | - α/α α |
| | The condition is called alpha thalassemia trait ;two α genes permit nearly normal production of red blood cells, but a mild microcytic hypochromic anemia is seen. The | |

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Lab Head-Clinical Genomics

| | | | |
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| | | |
|---|---|---|
| Two | disease in this form can be mistaken for iron-deficiency anemia and treated inappropriately with iron. Alpha-thalassemia trait can exist in two forms: * Alpha-thal-1(- -/α α), involves cis deletion of both alpha geneson the same chromosome. * Alpha-thal-2(- α/- α), involves trans deletion of alpha genes; this occurs on different (homologous) chromosomes | - -/α α or - α/- α |
| Three | This condition is called hemoglobin H disease; two unstable hemoglobins are present in the blood; hemoglobin Barts (tetrameric γ chains) and hemoglobin H (tetrameric β chains). Both of these unstable hemoglobins have a higher affinity for oxygen than normal hemoglobin. A microcytic hypochromic anemia with target cells and Heinz bodies (precipitated HbH) on the peripheral blood smear can occur, as well as hepatosplenomegaly. The disease is noticed in childhood or in earlyadult life; anemia and hepatosplenomegaly are noted | - -/- α |
| Four | This is known as alpha thalassemia major; these fetuses edematous, have little circulating hemoglobin, and the hemoglobin that is presentis all tetrameric γ chains.When all four alleles are affected, the fetus likely will not survive gestation without in utero intervention; most infants with alpha-thalassemiamajor are stillborn with hydrops fetalis. Fetuses treated with intrauterine transfusions throughout pregnancy starting at an early gestational age cansurvive to birth with acceptable morbidity. After birth, the treatmentoptions include bone marrow transplantation or continued chronic transfusions | - -/- |

αα/αα = normal: 'α α' before the '/' represents one chromosome, and 'α α' after the '/', its homologous chromosome.

*** End Of Report ***

**Disclaimer: Method given in report are only indicative and can be changed depending upon type of machine and kit available at time of testing.**

**Not all tests at all locations are under NABL scope. Availability of tests under NABL scope varies from lab to lab.**

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Booking Centre :- Hi Care Multispeciality Hospital (Ludhiana), Basnat Avenue, Near Bcm School
Processing Lab :- Redcliffe Lifetech Pvt. Ltd., H-55, Sector-63, Noida, Uttar Pradesh - 201301

# Terms and Conditions of Reporting

1. The presented findings in the Reports are intended solely for informational and interpretational purposes by the referring physician or other qualified medical professionals possessing a comprehensive understanding of reporting units, reference ranges, and technological limitations. The laboratory shall not be held liable for any interpretation or misinterpretation of the results, nor for any consequential or incidental damages arising from such interpretation.

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