


Name : Master. MOHAMMAD MUSTAKIN	Lab ID : 50600111016	CRM : 223003940862
Age : 5 Months	Sample Collection Date : 23-06-2025 16:00	
DOB : 26-12-2024	Sample Receipt Date : 25-06-2025 04:30	
Gender : Male	Reporting Date : 03-07-2025 11:08	
Referring Physician : Dr. SANDIP KARLE	Location : HINGOLI	
Hospital Name : BS20921		

HBB GENE SEQUENCING FOR MUTATION ANALYSIS

Details	Remarks
Sample Type	Peripheral venous blood
Quality of Sample	Adequate
Clinical Indication	To rule out Beta thalassemia status.
Test Requested	Beta Globinopathy

RESULTS

Variant Detected	Genotype	Allele Status	Clinical Significance
Common nomenclature: IVS-I-5(G>C) HGVS nomenclature: c.92+5G>C	β^*/β^*	Homozygous	Thalassemia Intermedia/ Major

INTERPRETATION

- A homozygous variant c.92+5G>C in intron 1 of the *HBB* gene, 5 base pairs downstream to the 5' splice site of exon 1 is detected in the **provided sample**. The identified mutation greatly reduces the efficiency of the 5' splicing site resulting in decrease in synthesis of the β Globin chain. According to HbVar database this variant is classified as β^* , and the disease indication is **Thalassemia Intermedia/Major**. The frequency of this variant among the affected population is found to be 46.55% in Maharashtrian and 30.5% in Punjabi population.

RECOMMENDATION

Kindly correlate the results with other clinical findings and Genetic counseling is recommended to understand the inheritance and risk of disease occurrence in future generations.

Methodology:

Sanger sequencing based DNA analysis was performed to identify the mutations in the coding regions and reported mutations in the non- coding regions of β Globin gene (HBB). DNA was isolated from the sample received using a commercial kit that works on the silica- membrane- based DNA purification. Mutation surveyor software V5.1.1 is used for identifying the mutations and the identified sequence alterations are

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This is an Electronically Authenticated Report.

Master. MOHAMMAD MUSTAKIN

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