# CHRISTIAN MEDICAL COLLEGE, VELLORE

## Department of Haematology

### Molecular Diagnosis Of Beta Thalassaemia (Seegene)

| Hospital Number | Patient Name | Gender | Age |
|---|---|---|---|
| 488062P | AQDAS TAHIR | MALE | 16 |

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|---|---|---|---|
| Date of Sample Collection : | 02/06/2023 | Referring Department : | HAEMATOLOGY -RANIPET |
| Lab Accession ID : | R-867 | Ordering Physician : | DR.THOLE HARSHIT PAWANKUMAR |
| Sample Type : | Peripheral Blood | | |

Report Date : 12/06/2023

Test Category : To screen for mutations in the β globin gene in the patient's DNA sample

Clinical Details : Frequency of transfusions:Once in 2weeks
Date of last transfusion:7 days back
Origin of family(ethnicity):Uttarpradesh
Provisional Diagnosis:Thalassaemia major

**Methodology**

Gene Tested : HBB haemoglobin,β. NCBI Reference Sequence: NC_000011.9NC_000011.9

Chromosome Location : 11p15

Method : Common mutations in the β-globin gene were screened for, using an in-house standardized NovaplexTM/ TOCE-BTA method. This method uses blood samples treated with a lysis buffer without DNA extraction for amplification and detection. Mutations are detected by DPO-TOCE-CMTA techniques. The tested mutations include 13 most common mutations; 12 point mutations: [HBB:c.27_28insG]/ Codon 8/9(+G), [HBB:c.47G>A]/ Codon 15(G>A), [HBB:c.92+1G>T]/ IVS I-1 (G>T), [HBB:c.92G>C]/ Codon 30(G>C), [HBB:c.92+5G>C]/ (IVS I-5(G>C), [HBB:c.126_129delCTTT]/ Codon 41/42(-TCTT), [HBB:c.79G>A]/ Codon 26 (G>A) (βE) and [HBB:c.20A>T]/ Codon 6(A>T) (HbS), [HBB:c.51delC]/ Codon 16 (-C), [HBB:c.46delT]/ Codon 15(-T), [HBB:c.-140C>T]/ -90(C>T), [HBB:c.-78A>C]/ -28(A>G), and 1 deletion: 619bp deletion. DNA was extracted from the patient by a commercial kit (Gentra purgene blood kit, Qiagen Gmbh). Mutations in beta globin gene were screened by bi-directional sanger sequencing.

Result : **Compound heterozygous for IVS I-5(G>C) [HBB:C>92+5G>C] and IVS I-129 (A>C) [HBB:c.93-2A>C] mutations.**

Impression : **Compound heterozygous for IVS I-5(G>C) [HBB:C>92+5G>C] and IVS I-129 (A>C) [HBB:c.93-2A>C] mutations in the beta globin gene. This genotype results into thalassaemia major phenotype.**

Note : Mutation nomenclature is as per the Human Genome Variation Society (HGVS).
TOCE-BTA has been developed and validated in-house
Other family members may be at risk of carrying this mutation and should be offered genetic counselling and testing.

Disclaimer : 1. This report should not be copied or reproduced except in its entirety
2. All precautions were taken to ensure the accuracy of these results. However, a 1% chance of error in this report is possible.

Reference : 1. www.hgvs.org
2. Hb Variant database (http://globin.cse.psu.edu/hb.var/menu.html)
3. Lee, YJ., Kim, D., Lee, K. et al. Single-channel multiplexing without melting curve analysis in real-time PCR. Sci Rep 4, 7439 (2014). https://doi.org/10.1038/srep07439

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