LABORATORY REPORT

Redcliffe labs

HBB GENE SEQUENCING ANALYSIS

Patient ID	NA	**Gender**	NA	**Location**	NA
Patient Name	Dummy	**Clinician Name**	NA	**Sample Collected**	DD-MM-YYYY
Patient DOB	NA	**GA/LMP Date**	NA	**Sample Received**	DD-MM-YYYY
Age	NA	**Hospital Name**	NA	**Report Released**	DD-MM-YYYY

Test Requested:- HBB Gene Sequencing **Sample Type:- NA** **Sample Quality:- Acceptable**

RESULT

Sample	Variant Detected	Genotype	Allele Status	Clinical Significance
Dummy	HBB: c.92+5G>C	IVS-1-5	No variant identified	Normal
	HBB: c.92+1G>T	IVS-1	No variant identified	
	HBB: c.27_28 insG	HbA	No variant identified	
	HBB: c. 125_128 CTTT del	Hb	No variant identified	
	HBB: 619 bp deletion	Hb	No variant identified	

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.

INTERPRETATION

No 5 common mutations is Identified in HBB Gene of Dummy.

TECHNOLOGY

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 reported in accordance with the Human Genome Variants Society (HGVS) nomenclature and annotated using one or more of the following database/s- ClinVar, OMIM, HGMD and SwissVar.

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UNDERSTANDING THE RESULTS:

Mutation Type	Possible Genotype*	Clinical Significance#
β+ type	β+ β+ β+ βo β+ β β β	**Thalassemia Intermedia / Major** **Thalassemia Major** **Thalassemia Minor** **Normal**
β0 type	βo βo β+ βo βo β β β	**Thalassemia Major** **Thalassemia Major** **Thalassemia Minor** **Normal**

***β type- Normal functional haemoglobin, β+ type Mutation: Reduced ability to produce β Globin protein with reduced functional capacity for hemoglobin, β0 type mutation: No β Globin will be produced and dysfunctional haemoglobin.**

#Thalassemia minor: Patient with one normal copy of the β-Globin gene and may show mild to no symptoms for Thalassemia, Thalassemia major: Patient with both abnormal copies of the β Globin gene and may show acute symptoms for Thalassemia, Thalassemia intermedia- Patient with both abnormal copies of the β Globin gene and may show acute symptoms for Thalassemia at adult stage.

DISCLAIMER:

This test does not distinguish between the alleles present in cis and trans forms. Classification of haemoglobin disorders by genetic data alone may result in incomplete conclusions which may significantly impact overall disorder classification and expected phenotype. The test results does not mean that the risk of carrying or developing Thalassemia/sickle cell anemia is not present and or caused by other known or unknown variations in the genome. Correlation with haemoglobin electrophoresis, red blood cell indices and clinical/family history is required. Limitations: This test will detect greater than 99% of variants in HBB gene excluding large insertions and deletions, which may be a rare phenomenon. Any change in primer binding site can result can interfere with the results and allele dropout cannot be ruled out using this experiment. Benign variants will be provided upon request. The current assay doesn't detect the delta beta inversion deletion mutation or any other mutation on Delta, Gamma and Alpha gene.

As per the PRE-NATAL DIAGNOSTIC TECHNIQUES (REGULATIONS & PREVENTION OF MISUSE) AMENDMENT ACT 2002, sex determination shall not be done for all prenatal samples

REFERENCE

Colah R, Gorakshakar A, Nadkarni A, Phanasgaonkar S, Surve R, Sawant P, Mohanty D, Ghosh K (2009) Regional heterogeneity of bthalassemia mutations in the multi ethnic Indian population. Blood Cells Mol Dis 42:241–246.

Edison ES, Shaji RV, Devi SG, Moses A, Viswabandhya A, Matthews V, George B, Srivastava A,Chandy M (2008) Analysis of b globin mutations in the Indian population: presence of rare and novel mutations and region-wise heterogeneity. ClinGenet 73:331–337.

http://globin.bx.psu.edu/hbvar/menu.html

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