Name	Master Soumyadip Naskar			Sample Type	Peripheral blood
Referred by	Dr. Sisir Patra			Patient ID	1002343232
Referring Centre	Dr. Sisir Patra - Kolkata				
Date Collected	22-06-2023	Date Received	24-06-2023	Date Reported	11-07-2023
Indication	HPLC report shows low HbA2 peak in the chromatogram				

HEMOGLOBINOPATHY MUTATION ANALYSIS REPORT (BY BETA GLOBIN GENE SEQUENCING)

Specimen Description: Peripheral blood: Specimen received was optimum for the test

Methodology:
HBB Gene sequencing (using Sanger sequencing)

PATIENT NAME	HGVS NOMENCLATURE	MUTATION	RESULT
Master Soumyadip Naskar	No point mutations detected in coding region of the HBB gene		

RESULT:

Master Soumyadip Naskar **:** No point mutations detected in coding region of the HBB gene

Sequencing Assay Limitations: *The assay detects mutations in the coding regions of the HBB (Human beta globin) gene. Mutations present in untranslated regions, large deletions eliminating coding sequence and mutations present in distal regulatory elements and genes associated with HBB gene will not be identified by this assay.*

Lilac Insights Pvt. Ltd. 301-302, Building A-1, Rupa Solitaire Millennium Business Park, MIDC Industrial Area, Sector-1, Navi Mumbai, Maharashtra 400710.
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Name	Master Soumyadip Naskar	Patient ID	1002343232

Beta-thalassemias are a group of hereditary blood disorders, characterized by decreased or absent synthesis of β-globin chains of hemoglobin resulting in variable phenotypes, ranging from severe transfusion dependent anemia to clinically asymptomatic individuals. In India, overall prevalence of Beta thalassemia carriers varies from 1.5% to 17% in different states. Beta thalassemia is caused due to mutations in the beta globin gene with more than 200 mutations reported globally. Five common mutations, IVS 1-5 (G-C), IVS 1-1 (G-T), 619 bp deletion (619 bpd), CD 8-9 (+G) and CD 41-42 (-TTCT) account for 80-85% of beta thalassemia carriers in India. The mutations are identified by ARMS-PCR. Carrier identification, genetic counseling and subsequent molecular diagnosis in high risk couples, aids in prenatal diagnosis of Beta thalassemia.
Genetic counseling is recommended for β-thalassemia, sickle cell &haemoglobinopathy carriers (traits).

DISCLAIMER:

This report is based on the sample received in the Lilac Insights laboratory; the analysis is based on the assumption that samples received are representative of the patient mentioned on the test requisition form and the sample. When samples are received from various referral centres, it is presumed that patient demographics are verified at the point of sample collection.
Chorionic villi samples yield DNA of lesser quality and quantity compared with other sample types. Despite all the necessary precautions and stringency adopted whilst performing DNA tests, the currently available data indicates that the technical error rate associated with all types of DNA analysis, is approximately 2%. It is important that all clinicians or persons requesting DNA diagnostic tests are aware of these data before acting upon these results. As with all diagnostic tests, the laboratory report must be interpreted in conjunction with the presenting clinical profile of the patient and evaluation of all reports.
In accordance to the Pre-Conception and Pre-Natal Diagnostic Testing (PCPNDT) Act, 1994- Govt. of India; Lilac Insights Pvt. Ltd. does not disclose the gender of the fetus.

LIMITATIONS:

Blood/Fetal samples may contain PCR-inhibitors which can inhibit DNA polymerases as well as primer annealing, preventing amplification of the target sequence and the consequence is that the mutation is not detected. PCR-ARMS can detect only known mutations and polymorphisms. For comprehensive mutation detection, PCR-ARMS should be combined with other mutation detection strategies like sequencing.

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Name	Master Soumyadip Naskar	Patient ID	1002343232

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