Name	Rootuja Anup Karhade		Age/Sex	29 yrs/F	Sample Type	Peripheral blood
Referred by	Dr. Chaitanya Shembekar				Patient ID	1002438370
Referring Centre	Shembekar Hospitals Pvt. Ltd.-Nagpur					
Date Collected	22-06-2024	Date Received	23-06-2024		Report Date	04-07-2024
Test confirmation	28-06-2024					
Indication	HPLC suggestive of sickle cell trait					

BETA THALASSEMIA MUTATION ANALYSIS REPORT

Specimen Description: Peripheral blood

Methodology:
Genomic DNA is isolated using standard protocol and the haemoglobinopathy HbS (CD 6 A-T) was screened using ARMS-PCR.

PATIENT NAME	HGVS NOMENCLATURE	RESULT	INTERPRETATION
Rootuja Anup Karhade	HBB:c.20A>T CD 6 (A-T)	Detected (Heterozygous)	Sickle cell trait

RESULT:

Rootuja Anup Karhade : Heterozygous for CD 6 (A-T): Sickle cell trait

Recommendation: Genetic Counselling is recommended.

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	Rootuja Anup Karhade	Age/Sex	29 yrs/F	Patient ID	1002438370

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 & haemoglobinopathycarriers (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.

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	Rootuja Anup Karhade	Age/Sex	29 yrs/F	Patient ID	1002438370

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-End of Report-