LILAC INSIGHTS PRIVATE LIMITED™
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CIN : U85191MH2011PTC217513

Name	Fetus of Mrs. Shabnam Khatun			Sample Type	Amniotic fluid
Referred by	Dr. Deepali Shukla			Patient ID	100204507
Referring Centre	Shukla Test Tube Baby Centre -Akola				
Date Collected	07-05-2020	Date Received	09-05-2020	Date Reported	18-05-2020
Indication	Previous child with beta thalassemia major				

BETA THALASSEMIA MUTATION ANALYSIS REPORT

Specimen Description: Parents: Peripheral blood
Fetus : Amniotic fluid: Specimen received was optimum for the test.

Methodology:
Genomic DNA is isolated using standard protocol and the common five β-thalassemia 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)] are detected using ARMS-PCR.
The mutations unidentified in the above panel are detected using automated DNA Sanger sequencing.

PATIENT NAME	HGVS NOMENCLATURE	MUTATION	RESULT	INTERPRETATION
Mr. Mohd. Imran	HBB:c.92+5G>C	IVS 1-5 (G-C) mutant & normal allele	Detected	Beta thalassemia minor
Mrs. Shabnam Khatun	HBB:c.92+5G>C	IVS 1-5 (G-C) mutant & normal allele	Detected	Beta thalassemia minor
Fetus	HBB:c.92+5G>C	IVS 1-5 (G-C) mutant allele	Detected	Beta thalassemia minor
	HBB:c.92+5G>C	IVS 1-5 (G-C) normal allele	Detected	

RESULT:

Fetus: Heterozygous for IVS 1-5 (G-C)

Hence the fetus is likely to be Beta thalassemia minor.

Parental mutations:

Mr. Mohd. Imran (Father): Heterozygous for IVS 1-5 (G-C): Beta thalassemia minor
Mrs. Shabnam Khatun (Mother): Heterozygous for IVS 1-5 (G-C): Beta thalassemia minor

No significant maternal contamination detected on PCR-based VNTR analysis with a lower limit of detection of 10%.

MKBanerjee

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Name	Fetus of Mrs. Shabnam Khatun	Patient ID	100204507

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.

Amniotic fluid 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.

REFERENCES:

Old JM, Varawaalla NY, Weatherall DJ. Rapid detection and prenatal diagnosis of β-thalassemia : studies in Indian and Cypriot populations in the UK. Lancet.1990;336:834-837

Vaz FE, Thakur CB, Banerjee MK, Gangal SG. Distribution of beta-thalassemia mutations in the Indian population referred to a diagnostic center. Hemoglobin. 2000;24:181- 94.

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