Laboratory Investigation Report

Patient Name	Centre
Age/Gender	OP/IP No/UHID
MaxID/Lab ID	Collection Date/Time
Ref By	Reporting Date/Time

SIN No:GGN275933

TEST REQUESTED

Sickle Cell Anemia Gene Mutation Analysis

METHOD USED

PCR, SEQUENCING

RESULTS

Mutation Not Detected

INTERPRETATION

Result	Comments
Homozygous Mutation Detected	Both alleles carry mutation
Heterozygous Mutation Detected	Single allele carries mutation
Not Detected	Both alleles do not carry mutation

NOTE

1. This is an in-house developed assay and detects both HbC c.19G>A, p.Glu7Lys and HbS c.20A>T, p.Glu7Val. Thus the following combinations are possible HbA/HbA, HbA/HbC, HbA/HbS, HbS/HbC, HbC/HbC or HbS/HbS.
2. Presence of PCR inhibitors in the sample may prevent DNA amplification.
3. Test can be conducted on whole blood for postnatal Mutation Analysis and Amniotic Fluid for Prenatal Mutation Analysis when parents are at high risk for hemoglobin abnormalities.
4. Genetic counselling is recommended.

COMMENTS

Sickle cell disease is caused by a mutation in the hemoglobin-Beta gene found on chromosome 11. Sickle cell disease is caused by one particular mutation on the HBB gene, producing an abnormal version of β-globin known as haemoglobin S (HbS) which can distort red blood cells into a sickle shape. Problems in sickle cell disease typically begin around 5 to 6 months of age. In sickle cell disease (SCD), some cells are curved -- like a sickle--and hard. Because they aren't as flexible--or deformable- they can get stuck in small blood vessels. This can happen in lots of organs like chest, belly and joints. More than 50% of people, experience acute chest syndrome because of the small vessels in the lungs. This can lead to acute hypoxia (low oxygen levels) as well as long term complications in the lungs. It is the stage known as sickle cell crisis.

(DR ATUL THATAI)
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Hematopathology

MC-2714

Test Performed at :910 - Max Hospital - Saket M S S H, Press Enclave Road, Mandir Marg, Saket, New Delhi, Delhi 110017
Booking Centre :5081 - Max Hospital - Gurugram, Opposite HUDA City Centre Metro Station, B - Block, 9911444185
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