



Laboratory Investigation Report

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

Molecular Diagnostics



SIN No: VSH1763019

Test Name	Result	Unit	Bio Ref Interval
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MTHFR Mutation Analysis*

Real Time PCR

MTHFR Mutation Analysis (RT-PCR)

MTHFR GENE, C677T	Not Detected
MTHFR GENE, A1298C	Heterozygous Mutation 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 qualitative assay.
2. All results should be interpreted in context of clinical findings.
3. This assay detects mutations in MTHFR gene (C677T & A1298C)
4. Test conducted on Whole blood.
5. Presence of PCR inhibitors if any, might lead to amplification failure.

Comments

Methylenetetrahydrofolate reductase (MTHFR) is an enzyme that breaks down the amino acid homocysteine. The *MTHFR* gene that codes for this enzyme can mutate, which can interfere with the enzyme's ability to function normally or completely inactivate it. Genetic polymorphism associated with severe MTHFR deficiency is defined by a C to T substitution at position 677 (C677T) and A to C substitution at position 1298 (A1298C) of the *MTHFR* gene. These mutations lead to the incorporation of amino acid alanine (A) instead of valine (V) at position 222 and glutamate to alanine substitution at codon 429 respectively of the MTHFR protein. These mutations may lead to hyperhomocysteinemia

Kindly correlate with clinical findings

*** End Of Report ***



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The authenticity of the report can be verified by scanning the Q R Code on top of the page

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Conditions of Reporting: 1. The tests are carried out in the lab with the presumption that the specimen belongs to the patient name as identified in the bill/test request form. 2. The test results relate specifically to the sample received in the lab and are presumed to have been generated and transported per specific instructions given by the physicians/laboratory. 3. The reported results are for the information and interpretation by the referring doctor only. 4. Some tests are referred to other laboratories to provide a wider test menu to the customer. 5. Max Healthcare shall in no event be liable for accidental damages loss, or destruction of specimen which is not attributable to any direct and mala fide act or omission of Max Healthcare or its employees. Liability of Max Healthcare for deficiency of services, or other errors and omissions shall be limited to fee paid by the patient for the relevant laboratory services.