

MTHFR (Methyl Tetrahydrofolate Reductase) gene mutation

MTHFR (Methyl Tetrahydrofolate Reductase) gene mutation (Real Time PCR)	Homozygous mutation detected
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Interpretation

RESULT	COMMENTS
Homozygous mutation detected	Both copies of the gene carry the mutation
Heterozygous mutation detected	One copy of the gene carries the mutation
Not Detected	Mutation not detected

Note:

1. This assay detects the mutation C677T in MTHFR.
2. This is an in-house developed assay
3. Test conducted on Whole blood
4. Genetic counseling available.

Comments

Homozygous mutation for MTHFR (Methyltetrahydrofolate reductase) is associated with hyperhomocysteinemia which is an independent risk factor for Stroke, Myocardial infarction, Peripheral arterial disease and venous thrombosis. Indian studies suggest that heterozygosity for MTHFR C677T is also associated with elevated homocysteine levels.

Uses

For evaluation of arterial and venous thrombosis risk