

Reference No.	: - 2411220318	Age/Gender	: 37 Yrs/Male
Pt's Name	: Mr. NAVPREET SINGH		AMB-KOS
Referred By	: NA		
Sample Collection Date/Time	: 14-Nov-2024	Date	:14-Nov-2024
Sample Receiving Date/Time	: 14-Nov-2024 05:38AM	Approved Date	:15-Nov-2024 06:37PM
Sample From	: KOS DIAG LAB	Report Print Time	:17-Nov-2024 07:02PM

Molecular Biology

Test Description	Observed Value	Biological Reference Interval
MTHFR Gene PCR*		
<u>MTHFR Mutation Detection</u>		
C677T*	Homozygous mutant	
A1298C*	Wild	

Method : Real Time Polymerase chain Reaction (PCR)

MTHFR (Methylene Tetrahydrofolate Reductase) is an enzyme involved in the anabolism of methionine, where it converts 5,10 methylenetetrahydrofolate to 5-methyltetrahydrofolate. This molecule, with homocysteine will form methionine in downstream processes. The gene for MTHFR expression is located on chromosome 1 of the human genome. Polymorphisms at 677 (C>T) and 1298 (A>C) reduce enzymatic activity. Reduction of MTHFR enzymatic activity results in increase of homocysteine levels and may produce hyperhomocysteinemia. Hyperhomocysteinemia is an independent risk factor for causing various blood vessel diseases including brain and/or heart blood vessel diseases and peripheral venous thrombosis. Because of the risks associated with reduction in MTHFR enzymatic activity, a method of determining the genetic status of the MTHFR gene is necessary.

Interpretation :

Limit of detection: 10 ng/ul

"Wild": No mutation in MTHFR gene

"Heterozygous mutant": Partial mutancy in MTHFR gene

"Homozygous mutant": 100% mutancy in MTHFR gene

Methodology details :

* DNA is extracted from samples by US FDA approved Automatic Extraction machine based on magnetic bead technology.

* Purified DNA is then Amplified and quantified using CE-IVD approved Real time PCR.

* Extraction and Amplification controls (IC) are incorporated in each run to ensure more accurate and precise detection of mutation.

*** End Of Report ***



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