

Lab Ref. No	:	Centre	: DR DANGS LAB
Name	:	Client	:
Age / Gender	: 34 Y 0 M 0 D /MALE	Collection Time	: 22-09-2023 11:59 AM
Ref. By Dr	:	Reporting Time	: 28-09-2023 04:18 PM
		Report Status	: Final Report

DEPARTMENT OF MOLECULAR BIOLOGY

TEST (METHODOLOGY)	RESULT	UNIT	BIO. REF. INTERVAL				
MTHFR (METHYLENE TETRAHYDROFOLATE REDUCTASE) GENE MUTATION C667T & A1298C, QUALITATIVE RT-PCR,							
Blood							

MTHFR (C677T) GENE POLYMORPHISM	NOT DETECTED	
MTHFR (A1298C) GENE POLYMORPHISM	NOT DETECTED	

INTERPRETATION:

• Methylenetetrahydrofolate reductase (MTHFR) is an enzyme that plays a central role in folate and homocysteine metabolism by catalyzing the conversion of 5,10-methylenetetrahydrofolate to 5-methyltetrahydrofolate, the bioactive form of folate.

• This enzyme is encoded by MTHFR gene which commonly shows genetic variants. Two of the most common genetic variants of the gene are - C677T and A1298C. These variants encode a thermo-labile form of the enzyme resulting in its deficiency of varying severity.

• Individuals who carry two copies of any of these variant (i.e homozygous for the variant), or 1 abnormal copy of 677C>T plus 1 abnormal copy of 1298A>C (double heterozygous) tend to have a lower enzymatic activity and higher homocysteine levels compared to those with wild type gene.

• The above testing is done by using a CE IVD approved Assay and detects these 2 common variants. Result should be interpreted in clinical context.

• MTHFR deficiency is the most common genetic cause of elevated levels of homocysteine in the plasma (hyper-homocysteinemia). Elevated plasma homocysteine is associated with increased incidence of cardiovascular events, stroke, dementia and Alzheimer's disease, increased incidence of bone fracture, and higher prevalence of chronic heart failure.

• Some studies have shown that women with homozygous form of these variants have a modestly increased risk of having a child with a neural tube defect and this risk increases if the fetus is also homozygous.

• Severe variants in the MTHFR gene can be a cause of an autosomal recessive inborn error of metabolism where extremely high levels of homocysteine accumulate in the urine and plasma. This can cause developmental delay, eye disorders, thrombosis, and osteoporosis.

*** End Of Report ***

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CONDITIONS OF REPORTING

- In case of alarming or unexpected test results you are advised to contact the laboratory immediately for further discussions and action. Laboratory results are meant to be correlated with the patient's clinical history.
- The report will carry the name and age provided at the time of registration.
- Reporting of tests will be as per defined laboratory turn around time for each test. The same will be informed to the patient during first point of contact i.e. registration or phlebotomy as the case may be.
- Test results & reference ranges vary depending on the technology and methodology used.
- Rarely a second sample may be requested for an indeterminate result or any other pre-analytical / analytical reason.
- Reports can be received either as a hard copy or an email on your personal ID. Reports can also be delivered via courier. Payments can be made online on our website. Only reports with no pending payments are mailed, uploaded or dispatched.
- Reports can also be accessed via Dr. Dangs lab website or through the Dr. Dangs mobile application on IOS and android using the unique ID and password provided to you during registration or received by you via SMS.
- Home collection sample facility is provided with prior appointment. Request for same to be given on 999-999-2020, booked online on www.drdangslab.com or through the Dr. Dangs mobile application on IOS and android.
- A digital invoice for tests performed is available on our website and can be accessed by using the unique I.D. and password provided.
- To maintain confidentiality, certain reports may not be mailed at the discretion of the management.
- In case of any queries pertaining to your test results or to provide feedback/suggestions please call us on 01145004200 or mail us at info@drdangslab.com.
- 48 hour notice is required for the issuing of slides and blocks.
- Test results are not valid for medico legal purposes.
- > The courts (forums) at Delhi shall have exclusive jurisdiction in all disputes/claims consuming the tests and/or results of the tests.
- *For any changes in timings, please visit our website.





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