Dr Test Doctor (Test Doctor) Test Clinic. 123 Test Street, Test Suburb Victoria 3125

Lab ID
Patient ID P000060
Ext ID 26115-0014

Test Patient

Sex: Female • 56yrs • 01-Jan-70
123 Home Street, Test Suburb VIC 3125

RECEIVED
25-Apr-26

Clinical Notes .

MTHFR GENOTYPE

Specimen type - Blood-EDTA

Collected
12-Nov-24

SERVICE	RESULT
MTHFR Gene A1298C	Negative
MTHFR Gene C677T	HETEROZYGOUS

Genetic Testing Comment

MTHFR Gene Mutation (A1298C): Negative - Mutation not found.

MTHFR Gene Mutation (C677T): **HETEROZYGOUS** for the mutation.

The patient has one copy of the MTHFR C677T mutation and is negative for the A1298C mutation. This is associated with intermediate enzyme activity (30-40% loss of function), but no increase in plasma homocysteine levels. MethyleneTetraHydroFolate Reductase (MTHFR) is a regulatory enzyme in folate-dependent homocysteine remethylation. A common polymorphism in the MTHFR gene at position 677 is associated with a thermolabile enzyme with decreased activity. The prevalence of the homozygous mutation ranges from 8- 18% in various populations. Clinically, homozygotes for the mutation have an increased risk of thromboembolism as well as premature vascular disease. A second mutation (A1298C) has also been described. This mutation is associated with an increased risk of thromboembolism, when only found together with the C677T mutation. Assessment of other biochemical markers involved in the methylation cycles will give a more in-depth assessment of the patient's methylation status/function. Assessments include Methylation Profile, Methionine Metabolism Profile, Folate Metabolism Profile, SAMe/SAH ratio, Homocysteine. Test performed by myDNA Laboratory NATA: 20082

Methodology

Quantitative PCR (qPCR)

SAMPLE REPORT

NATA Accreditation No. 20770. The current scope of accreditation is available on the NATA website.