NutriPATH
INTEGRATIVE PATHOLOGY SERVICES
P: 1300 688 522
E: info@nutripath.com.au

SAMPLE REPORT
09-May-1990 Female

16 HARKER STREET
BURWOOD VIC 3125

Dr.SAMPLE REPORT
TEST HEALTH CENTRE
123 TEST STREET
BURWOOD VIC 3125

LAB ID : 3814108
UR NO. :
Collection Date : 09-May-2022
Received Date:09-May-2022


3814108

GENOMIC_ASSESSMENTS

	Result	Range	Units

MTHFR Gene Mutation

MethyleneTetraHydroFolate Reductase (MTHFR) Gene Mutation.

MTHFR Gene Mutation (A1298C): HETEROZYGOUS for the mutation.
MTHFR Gene Mutation (C677T): HETEROZYGOUS for the mutation.

Method: Quantitative Real-time Polymerase Chain Reaction (qRT-PCR)

Comment:
The patient has one copy of the MTHFR C677T mutation and one copy of the A1298C mutation. This is associated with a 40 - 50% loss of function and an 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 accredited laboratory NATA: 20082

Tests ordered: MTHFR,IMPEI
(*) Result outside normal reference range (H) Result is above upper limit of reference rang