



HEATHER CATCHPOOLE
15-Jun-1983 **Female**

26 KINGSWAY
HAZELBROOK NSW 2779

P: 1300 688 522
E: info@nutripath.com.au

-.SUZANNE ELLIS
17 DAVID STREET
GLENBROOK NSW 2773

LAB ID : 4007573
UR NO. : 6291504
Collection Date : 24-Jul-2024
Received Date: 25-Jul-2024



4007573

Clinical Notes: Lamotrigine 150mg; Effexor 225mg;

GENOMIC_ASSESSMENTS

SWAB, Buccal Result Range Units

MTHFR Gene Mutation

MethyleneTetraHydroFolate Reductase (MTHFR) Gene Mutation.

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

MTHFR Gene Mutation (C677T): Negative - Mutation not found.

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

Comment:

The patient is negative for both the MTHFR C677T mutation and the A1298C mutation.

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

Tests ordered: MTHFR,GenomPEI