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-.NICOLE CHESTER HERB BAR 1/39 WAGAWN STREET TUGUN QLD 4224

HAYLEE MACKEN 20-Jan-1985 Female

27 PORTOBELLO DRIVE MERMAID WATERS QLD 4218

LAB ID: 4030074 UR NO.: 6307884 Collection Date: 02-Oct-2024 Received Date: 03-Oct-2024



4030074

GENOMIC ASSESSMENTS

SWAB, Buccal Result Range Units

MTHFR Gene Mutation

 ${\tt MethyleneTetraHydroFolate\ Reductase\ (MTHFR)\ Gene\ Mutation}.$

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

Tests ordered: GenomPEI,MTHFR