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26-Sep-1999 **Female**

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LAB ID : 4096149
UR NO. : 6368188
Collection Date : 29-Apr-2025
Received Date: 01-May-2025



4096149

GENOMIC_ASSESSMENTS

SWAB, Buccal Result Range Units

MTHFR Gene Mutation

MethyleneTetraHydroFolate Reductase (MTHFR) Gene Mutation.

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

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

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

Comment:

The patient has one copy of the MTHFR A1298C mutation and is negative for the C677T mutation. This is associated with minimal (20%) reduced enzyme activity and no increase in plasma homocysteine.

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