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LAB ID : 3833526
UR NO. : 6610380
Collection Date : 09-Aug-2022
Received Date: 11-Aug-2022

3833526

GENOMIC_ASSESSMENTS

SWAB, Buccal | Result | Range | Units

MTHFR Gene Mutation

MethyleneTetraHydroFolate Reductase (MTHFR) Gene Mutation.

MTHFR Gene Mutation (A1298C): HOMOZYGOUS for the mutation.
MTHFR Gene Mutation (C677T): Negative - Mutation not found.

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

Comment:
The patient has two copies of the MTHFR A1298C mutation.
This is associated with decreased enzyme activity (minimal up to 40% loss of function), but no increase in plasma homocysteine levels nor increased risk for venous thrombosis.

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