NutriPATH
Integrative Pathology Services
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TEST PATIENT
Sample Test Name
Sex : F
Date Collected : 00-00-0000
111 TEST ROAD TEST SUBURB
LAB ID: 00000000 UR#:0000000

TEST PHYSICIAN
DR JOHN DOE
111 CLINIC STREET
CLINIC SUBURB VIC 3000

GENOMIC_ASSESSMENTS

BUCCAL SWAB	Result	Range	Units

MTHFR Gene Mutation

MethyleneTetraHydroFolate Reductase (MTHFR) Gene Mutation.

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

Comment:
The patient has one copy of the MTHFR C677T mutation and one copy of the A1298C mutation.
This is associated with increased 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.

Tests ordered: IMPEI,CFee,MTHFR