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Date of Birth : 08-Jul-1987

Sex : F

Collected : 05-Aug-2016

27 BRINKLEY CIRCUIT

PALMERSTON ACT 2913

Lab id: **3440394** UR#:

PATIENT ADDRESS

GENOMIC_ASSESSMENTS

BLOOD SPOT

Result

Range

Units

MTHFR Gene Mutation, Bloodspot**MethyleneTetraHydroFolate Reductase (MTHFR) Gene Mutation.****MTHFR Gene Mutation (A1298C): Heterozygous for the mutation.****MTHFR Gene Mutation (C677T): 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.**