AUSTRALIAN Clinicalabs

LABORATORY 3427
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Referred: 07/06/16
Collected: 07/06/16 NS
Tested: 07/06/16
Printed: 23/06/16 12:00
Batch: 16254 4

461

MR PETER DONALD
TOTAL HEALTH CENTRAL COAST
37 TREELANDS DR
JILLIBY 2259

**MOLECULAR BIOLOGY** **SPECIMEN: BUCCAL SWAB**

**Methylenetetrahydrofolate (MTHFR)**

**MTHFR Gene Mutation (C677T) :** Mutation not found
**MTHFR Gene Mutation (A1298C):** Heterozygous for the mutation

**Comment:** The patient has one copy of the MTHFR A1298C mutation and is negative for the C677T mutation. This is not associated with increased plasma homocysteine nor venous thrombosis.

**Method:** Polymerase chain reaction (PCR) and sequence specific hybridisation.

**Clinical notes:** 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 been descibed. This mutation is associated with an increased risk of thromboembolism, only when found together with the C677T mutation.

**Reference:** Frosst P et al Nature Genetics 1995;10:111-13.
Weisberg IS et al Atherosclerosis 2001;2:409-15.

MFR-R


MTHFR-MTHFOLATE REDUCTA All Tests Complet

SURGERY USE: NORMAL ☐ NO ACTION ☐ CONTACT PATIENT ☐ SEE PATIENT ☐ FURTHER TESTS ☐