AUSTRALIAN Clinicalabs

LABORATORY 3427-3420 1868 DANDENONG ROAD CLAYTON VIC 3168 PH: 1300 554 480

PATIENT:	**REQUEST DETAILS:**	
RASHID VIC TEST	**LAB REF:** **17-4254154-MFF-0**	**TESTING SYSTEMS DEPARTMENT**
3 CHAPPEL ST	REFERRED: 01/03/17	IT DEPARTMENT
FOOTSCRAY VIC 3011	COLLECTED: 01/03/17 NS	1868 DANDENONG ROAD
PH: 95386742	REPORTED: 24/04/17 11:57	CLAYTON VIC 3168
DOB: 16/11/1989 SEX: FEMALE	TESTED: 01/03/17	
UR#: REF:	BATCH: 0 0	

MOLECULAR BIOLOGY **SPECIMEN: BUCCAL SWAB**

Methylenetetrahydrofolate (MTHFR)

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

Comment: This patient has one copy of the MTHFR A1298C and is clear for the C677T mutation. This is not associated with reduced enzyme activity nor increased plasma homocysteine levels.

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

Clinical notes: Methylenetetrahydrofolate reductase (MTHFR) is a regulatory enzyme in folate-dependent homocysteine remethylation. A mutation in the MTHFR gene may affect fertility in both sexes, mood disorders and mental health, autism spectrum disorders, risk of venous thrombosis and is associated with elevated plasma homocysteine. A common polymorphism (mutation)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. A second mutation (at position 1298) has been described and is associated with increased homocysteine levels when in association with the mutation at position 677.

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

MOLEC GENETICS

PATIENT: RASHID VIC TEST

ALL TESTS COMPLETE

PANEL CODES: MFF-R

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