



Lab ID: 394945220
UR: 394945220
MS CREMA, Tahlia
26 MILLER STREET
FITZROY NORTH VIC 3068

Patient tel#: 0421662184
DOB: 07/02/1987 SEX: F 36Y

M13384 Tg: POST Cr: PO

To: **DR CORNE MARE**
DOCTORS @ PACIFIC FAIR
PACIFIC FAIR SC, SHOP 1615B
CR GOLD COAST HWY & HOOKER BVD
BROADBEACH QLD 4218

MTHFR Genotyping

Specimen
MTHFR C677T
MTHFR A1298C

EDTA Blood
Heterozygous Mutation Detected
Heterozygous Mutation Detected

Please note these variants are known as c.665C>T, p.Ala222Val and c.1286A>C, p.Glu429Ala, according to HGVS nomenclature;
RefSeqGene: NM_005957.4.

This patient is heterozygous for both the thermolabile variant of the MTHFR gene (c.665C>T) and the A1298C variant (c.1286A>C). This combination of genotypes is unlikely to be clinically significant (Hickey et al., Genet Med 2013;15(2):153-156).

Test Information:

Testing was carried out by real-time PCR and melt curve analysis for two variants of the MTHFR gene, c.665C>T (p.Ala222Val, also known as C677T) and c.1286A>C (p.Glu429Ala, also known as A1298C).

NPAAC guidelines suggest that corroboration of genetic test results should be considered - e.g. by reference to other clinical or laboratory information or by repeat testing - as genetic tests may have medical implications for both the patient and their genetic relatives.

For clinician enquiries regarding these results, please contact Dr James Harraway (07 3377 8666).

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