

Lab ID 528650792

DOB 11/02/2005 (20 Yrs FEMALE)

Referrer To Racheal (Patient To Pay) Lee

Your ref.

Address COMPLEMENTARY MEDICINE 6/115 SHINGLEY BEACH DR
SHINGLEY BEACH RESORT
AIRLIE BEACH QLD 4802

Address 14/2 ERROMANGO DRIVE
JUBILEE POCKET QLD 4802

Phone

Phone 0497802078

Copy to To Commercial Testing Department (0733778485)

Requested 19/02/2025

Clinical Notes Not Provided

Collected 07/03/2025 10:49

Received 07/03/2025 10:55

MTHFR Genotyping

Specimen
MTHFR C677T
MTHFR A1298C

EDTA Blood
Heterozygous Mutation Detected *
Mutation Not 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.

Comments

This patient is heterozygous for the thermolabile variant of the MTHFR gene (c.665C>T). Heterozygosity for this variant is common, occurring in approximately 40-45% of the population. Heterozygosity for this variant has not been shown to be associated with increased risk of thrombotic disease.

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).

As they are typically only carried out once per patient, corroboration of genetic test results should be considered - e.g. by reference to other clinical or laboratory information or by repeat testing.
For clinician enquiries regarding these results, please contact Dr James Harraway (07 3377 8666).

HB

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Reported on 14-Mar-25 16:46