

BRITTANY BAXTER

Lab ID 528195107

DOB 20/01/1987 (38 Yrs FEMALE)

Referrer To Racheal (Patient To Pay) Lee

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Copy to

Requested 02/06/2025

Clinical Notes Not Provided

Collected 02/06/2025 13:48 Receiv

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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).

ER

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