

LEE, RACHEAL

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Patient **SPROUSTER, SIOBHAN KRISTINA**
 Patient Address **1-3 SHINGLEY DR AIRLIE BEACH QLD 4802**
 Sex **F** Age **30 years** DOB **15/06/1994**
 Report For **LEE, RACHEAL**
 Ref. by/copy to **LEE, RACHEAL**

UR No.

Requested 04/11/2024
Collected 04/11/2024 02:13 PM
Reported 07/11/2024 08:36 AM

MTHFR GENOTYPE ANALYSIS

SPECIMEN: Peripheral blood
REASON FOR REFERRAL: Nil stated

RESULTS:

C677T mutation: Heterozygous (single mutated allele detected)
A1298C mutation: NOT Detected

This patient is heterozygous for the MTHFR C677T mutation. Studies have shown that this mutation may result in intermediate enzyme activity, but is NOT associated with increase in plasma homocysteine levels or an increased risk of venous thrombosis.

METHOD:

The methylenetetrahydrofolate reductase gene (MTHFR, Ref. Sequence: NM_005957.4) was screened for the presence of C677T (also known as c.677C>T/c.665C>T) and A1298C (also known as c.1298A>C/c.1286A>C) mutations using real-time PCR analysis (Hanson et al. Clin Chem. 2001;661-666).

Note : Changes to MBS on 01/03/23 require a proven history of thromboembolism in the patient or a proven mutation in a first degree relative for the above test/s to be refundable by Medicare. As no relevant history was supplied/exists, the patient will be billed for these tests. Please contact Patient Accounts on 1800 350 046 to arrange appropriate billing if patient has a positive history.

For enquiries consult Dr Peter Davidson or Dr Abhijit Kulkarni.

Pathology Report