

LEE, RACHEAL

For Surgery Use ☐ Urgent ☐ Ring Patient ☐ Make Appointment ☐ Note in Chart ☐ File ☐

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|-----------------|---------------------|-------------------------------------|-----------|---------------------|
| Patient | BERRY, SHAINA ELYSE | 51 LITFINS RD MOUNT SYLVIA QLD 4343 | Requested | 25/07/2024 |
| Sex | F | Age 26 years DOB 07/11/1997 | Collected | 25/07/2024 02:26 PM |
| Report For | LEE, RACHEAL | | Reported | 30/07/2024 04:02 PM |
| Ref. by/copy to | LEE, RACHEAL | | | |

MTHFR GENOTYPE ANALYSIS

SPECIMEN: Peripheral blood
REASON FOR REFERRAL: Nil stated

RESULTS:
C677T mutation: Heterozygous (single mutated allele detected)
A1298C mutation: Heterozygous (single mutated allele detected)

This patient is heterozygous for the MTHFR C677T and A1298C mutations. Studies have shown that these mutations may result in increased plasma homocysteine levels, and a risk factor for 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