

LEE, RACHEAL

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

Patient **BOYLE-ENGLISH, JESSICA**  
 Patient Address **26 GILBERT ST BOWEN QLD 4805**  
 Sex **F** Age **30 years** DOB **05/12/1993**  
 Report For **LEE, RACHEAL**  
 Ref. by/copy to **LEE, RACHEAL**

UR No.

**Requested** 25/11/2024  
**Collected** 25/11/2024 12:00 PM  
**Reported** 04/12/2024 08:19 AM

## MTHFR GENOTYPE ANALYSIS

**SPECIMEN:** Peripheral blood  
**REASON FOR REFERRAL:** Nil stated

### RESULTS:

**C677T mutation:** Homozygous (two mutant alleles detected)  
**A1298C mutation:** NOT Detected

This patient is homozygous for the MTHFR C677T mutation. Studies have shown that this mutation may result in increased plasma homocysteine levels and an increased risk 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