

LEE, RACHEAL

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

Patient	POOK, MELANNIE LEE		UR No.
Patient Address	33 JUBILEE POCKET RD JUBILEE POCKET QLD 4802		
Sex	F	Age	49 years
DOB	27/01/1975		
Report For	LEE, RACHEAL		Requested 09/10/2024
Ref. by/copy to	LEE, RACHEAL		Collected 17/10/2024 11:46 AM
			Reported 29/10/2024 08:54 AM

## MTHFR GENOTYPE ANALYSIS

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

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

This patient is heterozygous for the MTHFR A1298C mutation. Studies have shown that without the presence of the C677T mutation, this is NOT associated with increased plasma homocysteine levels or 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

QML RTE001-AV4