

Pathology Report

LEE, RACHEAL

Urgent ☐ Ring Patient ☐ Make Appointment ☐ Note in Chart ☐ File ☐

Patient LIGHT, MICHAELA

Patient Address 116 MOUNT WHITSUNDAY DR AIRLIE BEACH QLD 4802

DOB 05/01/1998

Age 26 years

29/10/2024 Requested

Collected Reported

29/10/2024

07/11/2024

UR No.

08:07 AM 08:37 AM

Report For LEE, RACHEAL LEE, RACHEAL Ref. by/copy to

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.

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