

LEE, RACHEAL

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

Patient **LUCAS, EHELIA**

UR No.

Patient Address **8 EIGHTH CL BOWEN QLD 4805**

Sex **F** Age **31 years** DOB **22/01/1993**

Report For **LEE, RACHEAL**

Ref. by/copy to **LEE, RACHEAL**

| | | |
|------------------|------------|----------|
| Requested | 13/09/2024 | |
| Collected | 13/09/2024 | 08:55 AM |
| Reported | 25/09/2024 | 11:43 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

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CUMULATIVE SERUM HOMOCYSTEINE

Date 13/09/24
Time 08:55
Lab No 72509247

Homocysteine 10.4 umol/L (0.0-15.0)

72509247 High normal value.
With this level, the heterozygous state for defects of transsulphuration (homocysteinaemia) is unlikely. However the risk of coronary artery disease may be mildly elevated over the baseline. This is independent of other risk factors.

Homocysteine Related Risk

| Plasma level (umol/L) | Risk Average |
|-----------------------|--------------|
| Below 9.0 | No increase |
| 9.0 - 14.9 | x 2 |
| 15.0 - 19.9 | x 3 |
| 20.0 or greater | x 4.5 |

Risks approximated from New Eng J Med 1997 (337:230-236)

Pathology Report