Healius Pathology Pty Ltd (ABN 84 007 190 043)
APA No. 000042 t/a QML Pathology
11 Riverview Place, Metroplex on Gateway, Murarrie Qld 4172
Ph (07) 3121 4444

Pathology Report

LEE, RACHEAL

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

Patient **RILEY, KATE** **10 11 MAEVA ST JUBILEE POCKET QLD 4802**
Sex F Age 30 years DOB 24/04/1994
Report For LEE, RACHEAL
Ref. by/copy to LEE, RACHEAL

Requested 04/07/2024
Collected 04/07/2024 09:11 AM
Reported 09/07/2024 12:05 PM

Pathology Report

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.

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CUMULATIVE SERUM HOMOCYSTEINE

Date	04/07/24
Time	09:11
Lab No	76195731

Homocysteine	15.0 umol/L (0.0-15.0)

76195731 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

QML_ RTE001-AV3

Dept | Date of Service | For Clinical Enquiries | Perf. Branch | Quote 24-76195731