

LEE, RACHEAL

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

Patient **PALANCA, ANGELA FAYE**

UR No.

Patient Address **6 WANDERER AVE MERMAID WATERS QLD 4218**

Sex **F** Age **31 years** DOB **08/05/1992**

Requested 24/08/2023

Report For **LEE, RACHEAL**

Collected 24/08/2023 10:48 AM

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

Reported 01/09/2023 04:41 PM

MTHFR GENOTYPE ANALYSIS

SPECIMEN: Peripheral blood
 REASON FOR REFERRAL: Nil stated

RESULTS:

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

This patient is heterozygous for the MTHFR C677T mutation. Studies have shown that this mutation may result in intermediate enzyme activity, but is NOT associated with increase in plasma homocysteine levels or an increased risk of 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 01/05/00 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 or Genetics Dept on 07 3121 4466 to arrange appropriate billing if patient has a positive history.

For enquiries consult Dr Peter Davidson or Dr Kym Mina.

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Serum Reverse T3 (RT3) 379 pmol/L (170-539)

Pathology Report

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ADRENAL STUDIES

Serum Cortisol	220	nmol/L	(160 - 520)
Collection time:	10:48 am		

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CUMULATIVE SERUM HOMOCYSTEINE

Date 24/08/23
 Time 10:48
 Lab No 72998692

Homocysteine 9.7 umol/L (0.0-15.0)

72998692 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)