

MCEWEN, ELYSIA  
56 HILLCREST RD, QUAKERS HILL. 2763  
Phone: 0403753048  
Birthdate: 16/01/1987 Sex: F Medicare Number: 26101204061  
Your Reference: Lab Reference: 25-14800104-MTH-0  
Laboratory: 4Cyte Pathology  
Addressee: Dr ILONA SZEKELY Referred by: Dr ILONA SZEKELY

Name of Test: Mthfr Gene Test  
Requested: 22/01/2025 Collected: 29/01/2025 Reported: 19/02/2025 12:05

Clinical Notes: nil

#### **MTHFR Gene Testing (Whole Blood)**

MTHFR (C677T) Positive - Heterozygous variant identified  
MTHFR (A1298C) Positive - Heterozygous variant identified

This assay detects two of the most common variants in the methylenetetrahydrofolate reductase (MTHFR) gene C677T (also referred to as c.665C>T or p.Ala222Val) and A1298C (also referred to as c.1286A>C or p.Glu429Ala). These sequence variations are commonly known as single nucleotide polymorphisms (SNPs).

#### **Methods:**

Real-time PCR using TaqMan allele-specific primers and probes from Applied Biosystems.

Note: NATA accreditation does not cover the performance of this service. NSWHP and the RCPA do not recommend MTHFR testing for the above SNPs in view of the absence of an evidence base supporting clinical utility of testing. Further information regarding MTHFR genetic testing can be found on the RCPA website.

Test Performed at NSW Health Pathology John Hunter Hospital Laboratory.  
NATA/RCPA Accreditation Number: 3561.

Tests to follow: All Tests now completed

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Addressee: Dr ILONA SZEKELY Referred by: Dr ILONA SZEKELY

Name of Test: Mthfr Gene Test  
Requested: 22/01/2025 Collected: 29/01/2025 Reported: 12/02/2025 16:27

Clinical Notes: nil

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MTHFR (C677T) Positive - Heterozygous variant identified  
MTHFR (A1298C) Positive - Heterozygous variant identified

This assay detects two of the most common variants in the methylenetetrahydrofolate reductase (MTHFR) gene C677T (also referred to as c.665C>T or p.Ala222Val) and A1298C (also referred to as c.1286A>C or p.Glu429Ala). These sequence variations are commonly known as single nucleotide polymorphisms (SNPs).

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