NutriPATH
INTEGRATIVE PATHOLOGY SERVICES
P: 1300 688 522
E: info@nutripath.com.au

# ERIN JOHNSON
## 09-May-2000 Female

**97 BUENA VISTA ROAD**
**WINMALEE NSW 2777**

**-.SUZANNE ELLIS**
**17 DAVID STREET**
**GLENBROOK NSW 2773**

**LAB ID :** 4106057
**UR NO. :** 6358816
**Collection Date :** 21-May-2025
**Received Date:** 28-May-2025

4106057

**Clinical Notes:** Fluoxetine 80mg day

## GENOMIC_ASSESSMENTS

| SWAB, Buccal | Result | Range | Units |
|---|---|---|---|

### MTHFR Gene Mutation

**MethyleneTetraHydroFolate Reductase (MTHFR) Gene Mutation.**

**MTHFR Gene Mutation (A1298C): Negative – Mutation not found.**
**MTHFR Gene Mutation (C677T): HETEROZYGOUS for the mutation.**

**Method: Quantitative Real-time Polymerase Chain Reaction (qRT-PCR)**

**Comment:**
The patient has one copy of the MTHFR C677T mutation and is negative for the A1298C mutation. This is associated with intermediate enzyme activity (30-40% loss of function), but no increase in plasma homocysteine levels.

MethyleneTetraHydroFolate Reductase (MTHFR) is a regulatory enzyme in folate-dependent homocysteine remethylation.
A common polymorphism in the MTHFR gene at position 677 is associated with a thermolabile enzyme with decreased activity. The prevalence of the homozygous mutation ranges from 8- 18% in various populations.
Clinically, homozygotes for the mutation have an increased risk of thromboembolism as well as premature vascular disease.
A second mutation (A1298C) has also been described. This mutation is associated with an increased risk of thromboembolism, when only found together with the C677T mutation.

**Assessment of other biochemical markers involved in the methylation cycles will give a more in-depth assessment of the patient's methylation status/function.**

**Assessments include Methylation Profile, Methionine Metabolism Profile, Folate Metabolism Profile, SAMe/SAH ratio, Homocysteine.**

Test performed by myDNA Laboratory NATA: 20082

Tests ordered: MTHFR,GenomPEI