



MTHFR - Test code 5018



Turnaround Time: 7 Business Days



Specimen Type: Buccal swab or 1x EDTA Blood

Description

The MTHFR (methylenetetrahydrofolate reductase) gene test is a blood or buccal swab analysis that identifies genetic variations in the MTHFR gene, which plays a crucial role in the body's methylation process. This process is essential for converting folate into its active form, which helps in the production of DNA, the regulation of homocysteine levels and the detoxification of harmful substances. Variations in the MTHFR gene can reduce enzyme activity, potentially leading to elevated homocysteine levels, a recognised risk factor for cardiovascular disease, stroke and pregnancy complications (Raghubeer & Matsha, 2021).

Individuals with certain MTHFR variants may experience fatigue, mood changes or an increased risk of chronic conditions. This test is particularly valuable for patients with a family history of cardiovascular issues, recurrent pregnancy loss or unexplained fatigue (Raghubeer & Matsha, 2021).

Identifying MTHFR mutations can help healthcare providers personalise treatment strategies, including dietary adjustments and supplementation with activated forms of folate and B vitamins to support optimal methylation and homocysteine management (Raghubeer & Matsha, 2021).

Whats included?

- MTHFR (methylenetetrahydrofolate reductase) gene

Conditions and Symptoms

- Cardiovascular disease
- Depression and Anxiety
- Neural tube and other birth defects
- Elevated homocysteine
- Memory loss
- Reduce lead body mass

Complementary Testing

- Methylation profile (Test code 5101)
- Vitamin B12 & Folate (Test code 6013)

Accreditations Include:

- NATA ISO 15189 – Requirements for Quality and Competence in Medical Laboratories*
- CLIA – Clinical Laboratories Improvement Amendments*



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*See NATA and CLIA website for further details