



The MTHFR Gene

The MTHFR gene, technically referred to as Methylene tetrahydrofolate reductase, is a key enzyme required to metabolise homocysteine. Mutations of the MTHFR gene may cause elevated blood levels of homocysteine.

The most common mutation in the MTHFR gene is called C677T. Individuals with two copies of this mutation, i.e. one inherited from their mother and one from their father, are called homozygotes. This occurs in 5-10% of the population and these individuals are predisposed to developing high blood levels of homocysteine, particularly when their diets are low in folate.

A second mutation in the MTHFR gene, called A1298C, has also been implicated in high blood levels of homocysteine when found in conjunction with the C677T mutation.

High blood levels of homocysteine are recognised as a risk factor for:

- *Coronary artery disease*
- *Venous thrombosis & stroke*
- *Type 2 diabetes*
- *Obesity*

High homocysteine levels in the blood have also been associated with:

- *Neural tube defects*
- *Recurrent miscarriage*
- *Autism spectrum disorders*
- *Stillbirths*
- *Depression & other mood disorders*

The main causes of high homocysteine levels are folate deficiency, insufficient B12 and genetic mutations in the MTHFR gene.

Specimen Requirements

- *This test can be performed on either a blood specimen or buccal swab.*

Test Kit/Specimen Collection

Buccal swab – Once the practitioner has given the patient their request form, the patient can order their test kit online at www.functionalpathology.com.au or by calling Healthscope Functional Pathology customer service on 1300 55 44 80 between the hours of 8.30am and 5.30pm AEST. The test kit contains full instructions.

Blood - Once the practitioner has given the patient their request form, the patient takes it to their nearest Healthscope pathology collection centre. Please call 1300 55 44 80 or visit www.functionalpathology.com.au for a list of collection centres. Note that the blood specimen can be taken at any time of day and fasting is not required beforehand.

Turnaround Time

The standard turnaround time for this test is 7 – 10 working days from the date the patient's specimen/s are received by our laboratory.

Test Results

Patient results are delivered via mail, unless requested otherwise. Results can also be issued via:

- Fax
- Electronic Download
- Web Based Results

Technical Support

All Healthscope Functional Pathology tests are accompanied by an Interpretive Guide to assist

practitioners in their clinical understanding and patient management for each result. Healthscope Functional Pathology also has experienced full time Technical Advisors available for practitioners to discuss appropriate test selection, interpretation of test results, individual cases and other technical matters. Please call 1300 55 44 80 between the hours of 8.30am and 5.30pm AEST or email infofp@healthscope.com.au

Companion Tests

- **Homocysteine**
- **Red Cell Folate**
- **Vitamin B12**
- **2 & 16 Urinary Oestrogen Metabolites**
- **Baseline Hormone Profile**

A number of Healthscope Functional Pathology tests may be useful in conjunction with the MTHFR gene test. For example, given that folate deficiency is the major cause of high homocysteine levels, it is imperative to test Red Cell Folate to determine the level of insufficiency.

It is also recommended to test the blood levels of homocysteine and vitamin B12 which play an important role in the methylation cycle that converts methionine to homocysteine and eliminates it from the body.

The results of the MTHFR gene test in conjunction with the patient history and presenting complaints may also indicate the need for further testing to assess the whole picture. This may include the 2 & 16 Urinary Oestrogen Metabolites and Baseline Hormone Profile, for example, in female patients who present with depression and/or other mood disorders, obesity and personal or family history of cardiovascular disorders or fertility issues.