MTHFR Gene Testing

The MTHFR gene, technically referred to as Methylenetetrahydrofolate 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.

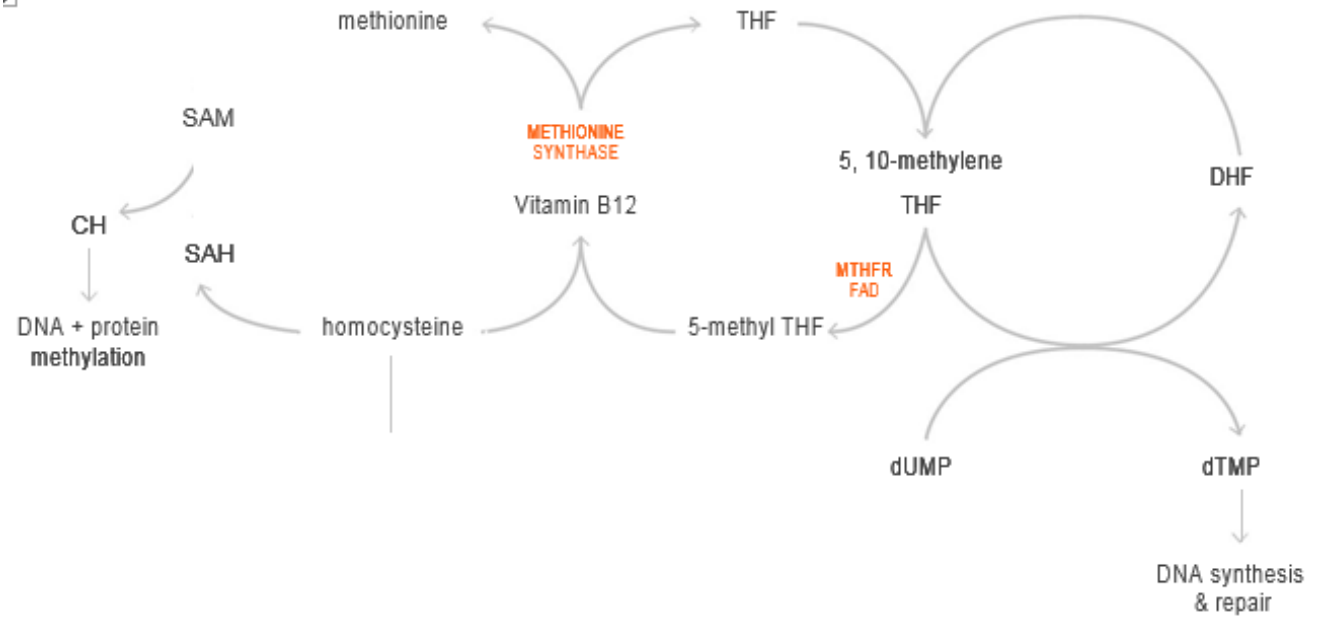
Individuals with two copies of this mutation, i.e. one inherited from their mother and one from their father, are called homozygotes. This occurs in 5-10% of the population and these individuals are predisposed to developing high blood levels of homocysteine, particularly when their diets are low in 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. The main causes of high homocysteine levels are folate deficiency, insufficient B12 and genetic mutations in the MTHFR gene.

High Homocysteine Risk Factors

- Coronary artery disease
- Venous thrombosis & stroke
- Type 2 diabetes
- Obesity
- Neural tube defects
- Recurrent miscarriage
- Autism spectrum disorders
- Stillbirths
- Depression & other mood disorders

Figure 1: The Methylation Cycle - Simplified



Source: Crott et al 2001

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 or by calling Australian Clinical Labs Functional Pathology customer service on 1300 55 44 80 between the hours of 9:00am and 5:00pm 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 Australian Clinical Labs pathology collection centre. Please call 1300 55 44 80 or visit www.clinicallabs.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.

Patient Preparation

- Patients must fast from 10pm the evening before the morning saliva specimen is taken (water may be consumed during this time)
- Patients must avoid all food and drink (except water) 30 minutes prior to collection of each specimen during the day
- Patients using hormone creams/gels/ patches must wait at least 12 hours after the last application before collecting their saliva specimen
- Patients using troches must wait three (3) days before collecting their saliva specimen

Turnaround Time

The standard turnaround time for this test is 10 – 14 working days from the date the patient's specimen/s are received at our laboratory.

Test Results

- Patient results will be delivered via electronic download, unless requested otherwise. However, we can also issue results via fax, hardcopy or web based e-viewer.

Technical Support

All Australian Clinical Labs Functional Pathology tests are accompanied by an Interpretive Guide to assist practitioners in their clinical understanding and patient management for each result. Australian Clinical Labs 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 9:00am and 5:00pm AEST or email csfp@AustralianClinicalLabs.com.au

Companion Tests

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

A number of 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.