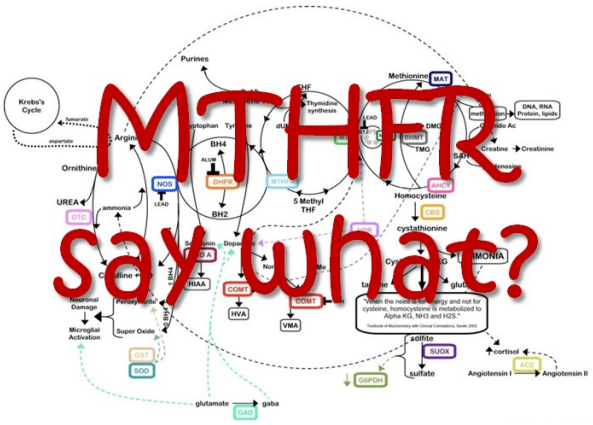


# MTHFR Gene Test

MTHFR is short for the enzyme, methylenetetrahydrofolate reductase. The MTHFR gene codes for this important enzyme that is involved in the conversion of folate (which is inactive) into its active form. The MTHFR enzyme affects this at the last step of conversion. So if you have a mutation in the gene then its going to be affecting how much active folate you are able to make.

**Folate is required for the following:**

- Synthesis of DNA, RNA and SAMe.
- Methylation: various critical reactions in the body require the donation of methyl groups to become active.
- Amino acid metabolism (for neurotransmitter, serotonin, dopamine and norepinephrine production and detoxification).
- Formation and maturation of RBC (red blood cells), WBC (white blood cells) and platelet production.
- Essential for detoxification of homocysteine.

**For all the above functions to happen the body needs to convert folate to the active 5-MTHF (5-methyltetrahydrofolate) form with the help of co-factors.**

Folate is required for methylation to work. Methyl groups (made up of one carbon and 3 hydrogen atoms) are required for a huge range of functions in the body. Methylation is the addition of this methyl group to another molecule. **If you are short of methyl groups many essential bodily processes cannot work properly and can cause a number of health related conditions.**

* Cardiovascular disease
* Anxiety
* Insomnia
* Fatigue
* Depression
* Autism
* Neural tube defects
* Preclampsia

The MTHFR gene test is used to detect two relatively common mutations in the *MTHFR* gene that are associated with **elevated levels of homocysteine** in the blood. People who have elevated homocysteine levels may be at increased risk of developing premature cardiovascular disease and/or thrombosis. ***MTHFR C677T*** and ***A1298C*** gene mutations are the most common and the ones that individuals are typically tested for.

MTHFR C677T Heterozygous = 40% loss of function
MTHFR C677T Homozygous = 70% loss of function

Heterozygous = 1 copy of the gene from either parent
Homozygous = 1 copy of the gene from each parent

**MTHFR Testing**

MTHFR gene testing can be performed either by a blood test or for children, a swab from the inner cheek within the mouth can be used.

All Natural Advantage



***John Pietryka***
*Biomedical Naturopath*
*55 Marianne Way*
*MT WAVERLEY 3149*
*www.allnaturaladvantage.com.au*
*Phone: (03) 8802 7687*
*E-mail:pietryka@optusnet.com.au*

Taking the guesswork out of diagnosis

## What follow-up testing to consider if you test positive for an MTHFR mutation?

The following tests may be appropriate to help with management of issues arising from a MTHFT mutation.

- Homocysteine
- Red cell folate
- Vitamin B12
- 2 & 6 urinary oestrogen metabolites
- Salivary hormone profile
- Functional Liver Detoxification Profile (FLP)

## Further considerations if you have the MTHFR mutation.

### Do not take supplements with folic acid

- Individuals with the MTHFR mutation have a limited ability to convert folic acid into methyl-folate
- High levels of unmetabolised folic acid could be detrimental to an individuals health
- Folic acid blocks the folate-receptor sites that are used by methylfolate

### Take additional vitamin B12

- Individuals with MTHFR mutations are low in vitamin B12. Folate and vitamin B12 work together. Testing vitamin B12 levels are recommended prior to supplementation either by blood testing or urinary for methylmalonic acid (MMA)
- If supplementing with vitamin B12, check product label whether the supplement contains folic acid. Gene mutations in n.vitamin B12 metabolism may also increase the need for vitamin B12. Supplement with active forms of vitamin B12 (methylcobalamin, adenosylcobalamin or hydroxocobalamin)

### Take a daily active folate supplement

- Take an active form of folate such as l-methyltetrahydrofolate (L-MTHF)
- Begin with a low dose and under the supervision of a health practitioner increase the dose slowly
- **Caution:** there is some concern that excess folate may cause hypermethylation and promote cancer and ageing. Always seek the advice of a practitioner that understands the complexities of supplementing with folate supplements and to monitor treatment with appropriate testing. If taking pharmaceutical medications do not supplement without checking with your doctor or health practitioner for possible adverse interactions.

MTHFR is an important gene, but there are many other critical genes and single nucleotide polymorphisms (SNPs) that can also affect your wellbeing. Consider a personal DNA profile to identify your genetic weaknesses that may be affecting your health.