

## What Is MTHFR?

### What is folate?

Folate is Vitamin B9 and is NOT the same as folic acid. (Folic acid is synthetic and is not found in nature. It must undergo various transformations to utilization.)

Folate is required for the following:

- Synthesis of nucleic acids (for DNA production and repair and tRNA)
- Single carbon metabolism (methylation)
- Interconversion of amino acids (for neurotransmitter production and detoxification)
- Formation and maturation of RBC, WBC 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) with the help of co-factors.

This process is known as methylation. Many factors can affect methylation, however the MTHFR gene mutation is a critical one.

### What is MTHFR?

MTHFR is a gene, which produces the methylenetetrahydrofolate reductase (MTHFR) enzyme. This produces the final form of folate 5-MTHF and provides the substrate for the transmethylation cycle and is found inside each cell.

### The MTHFR Genes

SNPs that we currently test for:

1. MTHFR C677T
2. MTHFR A1298C

Heterozygous = 1 copy of the gene from either parent

Homozygous = 1 copy of the gene from each parent (most commonly – some exceptions)

MTHFR C677T Heterozygous = 40% loss of function

MTHFR C677T Homozygous = 70% loss of function

MTHFR A1298C Heterozygous = (loss of function - research not known)

MTHFR A1298C Homozygous = between 50-70%

MTHFR C677T & MTHFR A1298C heterozygous = compound heterozygous = 50% loss of function

### What happens if MTHFR activity is reduced?

1. 5-MTHF levels go down

2. All tissues except liver and kidney show effects of decreased methylation which leads to:

- Decreased neurotransmitter production
- Deficiency in glutathione

3. BHMT becomes depleted in choline which causes:

- Homocysteine levels to rise
- Methylation in liver and kidneys decrease
- Phosphatidylcholine production drops causing cell membrane and myelin instability causing neuropathy and parkinsons type conditions

This will then have affects for the following conditions:

- ADD/ADHD
- Addictive behaviours
- Alcoholism
- Allergies
- Alzheimers
- Anxiety
- Autism
- Autism
- Bipolar
- Cancer
- Cardiovascular disease – Heart attack, atherosclerosis, elevated cholesterol, hypertension, stroke, fat metabolism issues
- Chemical sensitivity
- Chronic fatigue syndrome
- Chronic viral infection
- Cleft palette
- Congenital Heart defects
- Depression
- Diabetes
- Downs syndrome
- Fibromyalgia
- Immune deficiency
- Infertility
- Insomnia
- MS (multiple sclerosis)
- Neural tube defects
- Neuropathy
- Nutritional disorders (Vitamin & mineral imbalances that lead to disease)
- Panic attacks
- Parkinsons
- Pulmonary embolisms
- Recurrent miscarriage
- Schizophrenia
- Spina bifida
- Thyroid dysfunction

### **Current MTHFR testing**

Testing for the gene MTHFR can be done in pathology or with a home test kit, see more information about testing here:

<http://mthfrgenehealth.com/mthfr-testing/>

### **What to do once you are confirmed with the mutation in the MTHFR gene.**

1. Avoid all sources of folic acid. If homozygous to C677T , that includes commercial bread which is fortified with folic acid.

2. Seek professional advice as to how to bring the methylation cycle back into balance here:

<http://mthfrgenehealth.com/mthfr-treatments/>

MTHFR is just a small part of this cycle and although restoring the deficiency due to MTHFR gene mutation may help most people, this may not be the solution for everyone.

3. Only the 5-MTHF active folate should be given after consultation with a MTHFR practitioner.

4. Avoid drugs that affect the methylation cycle:

- Antacids (deplete B12)
- Cholestyramine (deplete cobalamin and folate absorption)
- Colestipol (decrease cobalamin and folate absorption)
- Methotrexate (inhibits DHFR)
- Nitrous oxide (inactivates MS)
- High dose Niacin (depletes SAMe and limits pyridoxal kinase – active B6)
- Theophylline (limits pyridoxal kinase = active B6)
- Cyclosporin A (decreases renal function and increases Hcy)
- Metformin (decreases cobalamin absorption)
- Phenytoin (folate antagonist)
- Oral contraceptives ( deplete folate)
- Antimalarials JPC-2056, Pyrimethamine, Proguanil (inhibits DHFR)
- Antibiotic Trimethoprim (inhibits DHFR)
- Ethanol (depletes folate and cobalamin)
- Bactrim (inhibits DHFR)
- Sulfasalazine (inhibits DHFR)
- Triamterene (inhibits DHFR)

6. Avoid drugs that raise homocysteine levels.

A number of prescription drugs and natural compounds can elevate blood levels of homocysteine by interfering with folate absorption or metabolism of homocysteine. These include:

- **Caffeine** - Cafcit®, Cafergot®, Esgic®, Excedrin Migraine®, Fioricet, Fiorinal, Norgesic®, Synalgos-DC®
- **Cholestyramine** - Questran®, Questran Light®, Cholybar®
- **Colestipol** - Cholestid®
- **Fenofibrate** Antara®, Fenoglide®, Lipfen®, Lofibra®, Tricor®, Trilpix®
- **Levodopa** Parcopa®, Sinemet®, Stalevo®
- **Metformin** ActoPlus Met®, Avandamet®, Fortamet®, Glucophage®, Glucovance, Glumetza®, Janumet®, Metaglip®, Prandimet, Riomet®
- **Methotrexate** Rheumatex®
- **Niacin** Advicor®, OcuVite, CardioBasics®, CitraNatal®, Heplive®, Niaspan, Simcor®
- **Nitrous oxide**
- **Pemetrexed** Alimta®
- **Phenytoin** Dilantin®, Phenytek®
- **Pyrimethamine** Daraprim®, Fansidar®
- **Sulfasalazine** Asulfidine®

7. Ask questions, get informed, get educated by MTHFR practitioners here:

<http://mthfrgenehealth.com/contact-us/>