MTHF Defects

Jacqueline Fields, MD and founder of Dr. Fields' Sacred Skin| June 2016

Although there are over fifty known MTHFR polymorphisms that can affect the amount and activity of this vital enzyme, the two primary ones we need to think about are called C677T and A1298. These are the most common genetic variants that cause MTHFR to function at a lower than normal rate and thereby slow the methylation process in tissues throughout the body.

Methylation is a core process that occurs in all cells. It is responsible for:
Cellular Repair: synthesis of nucleic acids, production and repair of DNA and mRNA.
Detoxification and Neurotransmistter Production: interconversion of amino acids.
Healthy Immune System Function: formation and maturation of red blood cells, white blood cells & platelet production.

MTHFR-VitB12When people with mutations in MTHFR are exposed to environmental or food borne toxins, they have a harder time getting rid of them, which can cause some very serious illnesses.

The 677T variant is most commonly associated with early heart disease and stroke. The 1298C variant is linked to a wider variety of chronic illnesses, but either anomaly can cause health problems.

If someone is heterozygous for one of these, his or her MTHFR enzyme activity will run at about 60% efficiency compared to someone without these polymorphisms.
If someone is homozygous, then enzyme efficiency drops down to 10% to 20% of normal, which can be very serious.

The worst combination is 677T/1298C in which someone is heterozygous for both anomalies.

Many chronic illnesses are linked to this "double whammy." Fibromyalgia, irritable bowel syndrome, migraines, chemical sensitivity, frequent miscarriage and frequent blood clots are all conditions associated.

MTHFR-Related Health Problems

Glutathione is the body's primary antioxidant and detoxifier. One of the ways that MTHFR gene mutation can increase susceptibility to illness is by compromising the ability to make glutathione. People with MTHFR anomalies usually have low glutathione, which makes them more susceptible to stress and less tolerant to toxic exposures.

Accumulation of toxins in the body and increased oxidative stress, which also leads to premature aging.

Some other conditions that may be associated with MTHFR gene mutations include:

Autism
Addictions: smoking, drugs, alcohol
Down's syndrome
Frequent miscarriages
Male and female infertility
Pulmonary embolism and other blood clots
Depression and anxiety
Schizophrenia
Bipolar disorder
Fibromyalgia
Chronic Fatigue Syndrome
Chemical Sensitivity
Parkinson's disease
Irritable Bowel Syndrome
Stroke
Spina bifida
Migraines
Hyperhomocysteinemia
Breast cancer
Atherosclerosis
Alzheimer's
Multiple Sclerosis
Myocardial Infarction (Heart Attack)
Methotrexate Toxicity
Nitrous Oxide Toxicity
Treating MTHFR Polymorphisms

It is easy to test for the most common and problematic MTHFR polymorphisms. Many functional diagnostic labs offer tests, and the information can be extracted from consumer-facing genomic tests such as 23andMe.

If someone has one or more of the problematic mutations, you can supplement with methylfolate and methyl B12, the active forms of these B vitamins. Some of my favorites are Thorne Research's Methyl Guard Plus and 5-MTHF 1mg and 5mg.

You can also supplement with liposomal or acetyl-glutathione, the end product of the MTHFR-mediated biochemical pathway. Glutathione is poorly absorbed, so either the liposomal form or a precursor, called n-acetylcysteine (NAC) may be used. Or we carry S-Acetyl Glutathione from xymogen or from a pharmacy. The data on these 2 options show great absorptability.

There are also prescription medications that contain methyl-folate: Deplin, MetanX, CerefolinNAC are a few. It's also possible to give methyl B12 via intramuscular injection, as a nasal spray, or sublingually.

The intramuscular shots are by far the most effective method, but have obvious limitations in terms of routine use and patient compliance.

Other B vitamins, such as riboflavin and vitamin B6 also play an important role in the care of patients with MTHFR polymorphisms. As you may have surmised, this can be quite complex, and it is important to keep in mind that it is not uncommon for patients with MTHFR polymorphisms to be very sensitive to supplementation.

Who Should Be Screened?

I recommend testing for MTHFR mutations in all patients with:

Mood disorders: depression, anxiety, irritability, mood swings, bipolar symptoms
Infants and children of parents with MTHFR mutations
Family members related to someone with MTHFR mutations
Infertility and Pre-conception care: test both man and woman
Elevated folate (not processing to active 5-MTHF due to inability to methylate)
Elevated homocysteine (due to low active 5-MTHF and methylcobalamin)
Elevated s-adenosylhomocysteine (due to low active 5-MTHF and methylcobalamin)
Elevated serum cobalamin (due to inability to methylate cyanocobalamin to methylcobalamin)
Elevated methylmalonic acid (due to methylcobalamin deficiency)
Patients with complex chronic "syndromes:" IBS, multiple chemical sensitivity, fibromyalgia, Down syndrome, chronic fatigue syndrome
Neurological disorders: Multiple sclerosis, Autism, Alzheimer's, Epilepsy, Parkinson's
Cancer: family history of cancer or undergoing cancer treatment
Cervical dysplasia
Cardiovascular risk: family history of strokes, embolisms, heart attacks, clots, hypertension
Birth defects: cleft palate, tetralogy of Fallot, spinal bifida, midline defects
Drug sensitivities: methotrexate, anti-seizure meds, nitrous oxide, anesthesia.
If you start looking for these polymorphisms, it's very likely you will find them. This will open new opportunities to minimize risk and truly optimize patients' health through safe and effective nutrition-based interventions.

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