# MTHFR Mutations

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For the past 2 years, I have tested almost every one of my patients for the MTHFR mutation. *(MTHFR = methylenetetrahydrofolate reductase - I will explain more about it as you read, so please don't give up yet.)* Over this time, I have found the majority of my patients (about 75%) have at least a single MTHFR mutation and upwards of 40% of my patients have a double mutation (either heterozygous (one of each) A1298C & C677T, homozygous (two) C677T or homozygous A1298C). The other doctors in my office also find similar percentages amongst their adult patients. Many research journals confirm similar frequencies, but often only test one or the other mutations. So this is a very common mutation and we have a lot of patients who want to understand more about it. So this post is for them, and maybe also for you too.

What does the MTHFR mutation do? Having a MTHFR mutation means that the enzyme that converts folic acid into it's activated form (5-MTHF) acts in a sluggish manner. Having two defects at the same point makes it more sluggish than just having one. As taught in basic chemistry, enzymes are the "rate limiting factor" to any biochemical reaction. Thus when an enzyme is sluggish, the biochemical reaction that relies upon it occurs slower than it should. When one pathway is working slower than others, the other pathways that depend upon it either back up or find a detour to shunt their excess product. Similar to what you may see on a highway, detour pathways are usually not as efficient, using more energy (ATP), and are prone to back ups, often causing new problems to be managed.

The MTHFR enzyme metabolizes folic acid into 5-MTHF, needed to combine with homocysteine to break it down and to facilitate methylation processes in the body. Thus, what is often seen with MTHFR mutations are: elevated homocysteine levels and defective methylation.* High homocysteine is associated with increased heart disease risk, strokes and blood clots. When the MTHFR mutation is correctly supported through vitamin supplementation (nutritionally bypassing the mutated enzyme and redirecting the pathway with B12 and sometimes P5P), homocysteine levels decrease, often quickly and dramatically, and patients see results clinically.

*[*Homocysteine levels will often be normal even in the face of homozygous MTHFR mutations. I see this occuring frequently in children, whose homocysteine levels typically are much lower than adults (homocysteines of 4-6) whereas normal adult levels are 8. Thus a normal homocysteine does not at all promise a normal MTHFR genetic profile. Homocysteine levels can be elevated in people who have B12 deficiency, as B12 is a cofactor in the breakdown of homocysteine. Thus high homocysteines do not promise MTHFR mutations either.]*

Methylation is required for many processes in the body, from breaking down histamine, seratonin and dopamine to turning DNA on and off. Consequently, having a defective methylation capability is associated with psychiatric illnesses, such as schizophrenia, depression and bipolar, as well as autoimmunity disorders, ADD, autism, spina bifida, Down's syndrome, miscarriages, and cancer. Defective methylation can come from not having the 5-MTHF or not having enough B12, so the disorders and symptoms associated with the MTHFR mutations often overlap with those due to B12 deficiency. Many may simultaneously have deficiencies in both.

As B12 helps to redirect the detour to the more efficient pathway, I always recommend that my patients with MTHFR mutations take both 5-MTHF and B12.

While I don't test every one of my patients for the MTHFR mutation, I certainly try to test everyone who has a family history of cardiovascular disease (heart disease, stroke, blood clots), mental illness, autoimmune disorders, spinal cord defects, cancer or miscarriages. As these disorders are all too common, this explains why I end up ordering MTHFR testing, as well as B12 levels, on the vast majority of my patients.

I find that knowing your MTHFR status is far more empowering than it is scary. Rather than keeping your head in the sand, it allows you to provide your body with what it needs to function most efficiently and hopefully prevent disease. Finding that you have a mutation, or even two, also encourages you to get your other family members tested. Parents and siblings may likely also carry this mutation and often benefit greatly from the additional knowledge and support.

To review MTHFR research, see Dr. Ben Lynch's site MTHFR.net where he has many listed.

*From Dr. Katherine Erlich's web site with permission*
*http://healingthewholechild.com/2012/03/01/mthfr-mutations/*