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MTHFR

(Methylenetetrahydrofolate reductase)

MTHFR is a common genetic variant that causes a key enzyme in the body to function at a lower than normal rate. This can lead to a variety of medical problems, when people with MTHFR are exposed to more toxins than their bodies can handle. There are over 50 known MTHFR variants, but the two prime variants are called 677 and 1298, the numbers refer to their location on the gene. The routine lab test for MTHFR variant only reports on 677T and 1298C as these are the most studied.

The 677T variant is most commonly associated with early heart disease and stroke and the 1298C variant with a variety chronic illnesses, but either anomaly can cause a wide variety of health problems. The MTHFR anomaly is reported out as heterozygous or homozygous. If you are heterozygous that means you have one affected gene and one normal gene. The MTHFR enzyme will run at about 55% to 70% efficiency compared to a normal MTHFR enzyme.

If you are homozygous then enzyme efficiency drops down to 7% to 10% of normal, which of course makes a huge difference. The worst combination is 677T/1298C in which you are heterozygous to both anomalies. Many chronic illnesses are linked to this anomaly. 98% of autistic children have an MTHFR anomaly. Fibromyalgia, irritable bowel syndrome, migraines, are all conditions associated with MTHFR anomaly. We have also rarely seen patients homozygous with 677T and heterozygous 1298C, but this is very rare.

MTHFR can make you susceptible to illness because the pathway is the primary source of glutathione production in the body. Glutathione is the body's primary antioxidant and detoxifier. People with MTHFR anomalies usually have low glutathione, which makes them more susceptible to stress and less tolerant to toxins. As we age MTHFR problems get much worse due to the accumulation of toxins and the cumulative effect of oxidative stress, which ages our bodies.

Treatment:

Fortunately, we can now easily test for MTHFR and augment the essential nutrients made by the MTHFR enzyme, which is methyl B12. the active form of B12 and glutathione the end product of the pathway. There are prescription medical foods that help: Deplin, MetanX, Nees, CerefolinNAC are a few of the compounds available. Methyl B12 can be given as shots, nasal sprays, and sublingually. The shots are by far the most effective method. There are other supplements, SAME, DMG, and Betaine that are useful in augmenting the methylation pathway. However, some of these compounds especially SAME may actually make things worse in some people. Biopterin commonly referred to as BH4 is another compound that can be very helpful especially in diabetics and chronic depression. The choice of nutrients will vary from patient to patient. Having adequate levels of B vitamins is important, since lack of B vitamins, especially vitamin B6 may cause problems in the pathway.

The choice of treatments is somewhat dependent on the nature of the anomaly. I have found in the 1298 anomaly methyl B12 works very well and in the 677 anomaly, L-methyl folate (Deplin, Metanx, some supplements) is most helpful. Although, I often use multiple compounds to augment the pathway at several points.

Once a MTHFR variation is confirmed then a number of steps can be taken to minimize health problems due to MTHFR. The treatment of MTHFR related problems is primarily nutritional. There are a number prescription medications, which are classified as medical foods, that are designed to help with this disorder. The two that I use the most are **Metanx** and **Deplin**. The links take you to the pharmaceutical websites, which do not discuss the full range of conditions for which these medications are useful. Treatment of patients with MTHFR related issues can be simple or complex depending on a variety of factors including: life style, diet, and overall accumulated toxicity. There are two primary issues: the first

is augmenting the pathway to produce adequate glutathione and the second is dealing with the accumulated toxic load. This second issue makes treating MTHFR related problems tricky. When glutathione levels are raised, the body will start excreting these accumulated toxins. This done primarily via the liver which excretes the toxins into bile. The bile is then excreted into the intestinal tract, where the toxins can be released and reabsorbed, which can cause a whole variety of nasty side effects. These side effects can be minimized by using compounds to bind up bile in the GI tract, such as Toxin Free, activated charcoal, modified fruit pectin, and cholestyramine, a drug used to bind bile. Many of these problems can be avoid by starting the treatment of the methylation pathway slowly. That is to start with smaller than maintenance doses and ramp up the pathway slowly.

Treatment Tip: an adverse reaction to treatment often means, you are on the right track, but need to change the dose or rate of treatment. The same nutrient given at lower dose may be very helpful. You may also need more B vitamins especially vitamin B6.

I have put together a couple of mind maps that will help you visualize the condition. There is also a link to information put together by Dr. Neil Rawlins including a number of his lectures on MTHFR. My mind maps closely follow his lecture notes. I wish to thank Dr Rawlins for all the work he done in this area. While MTHFR was something I knew about on an intellectual basis, his lectures made me realize that MTHFR was a primary cause of many of my patient's health problems.

My key recommendations to patients with chronic medical disorders are:

- Get evaluated for MTHFR. It is a simple blood test costing about \$160.00.
- Consult with a physician who knows how to treat MTHFR related problems.

The first link will take to Neil Rawlins's site, where you can download his series of lectures on MTHFR. A mind map is a way to present complex material in a graphic fashion. These maps are published on Mindmeister.com.

[Dr. Neil Rawlins' Lectures and Notes on MTHFR](#)

[MTHFR related medical conditions](#)

[MTHFR Treatment](#)