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The MTHFR Defect – Are You at Risk?

JUN  
 8 6/8/2011 8:20 AM



There is a good chance that most people reading this article do not know what the MTHFR gene is, and this is a problem. It's estimated that up to 50 percent of the population may have a defect in the MTHFR gene, and this defect can result in numerous health problems, including colon cancer and cardiovascular disease, and may be related to miscarriages and health and development of fetuses.

MTHFR is an acronym for *methylenetetrahydrofolate reductase*, and a problem with the gene is referred to as the MTHFR defect. This defect impairs the function of an enzyme that activates folic acid, commonly known as vitamin B9. Vitamin B9 is involved with cell growth and replication, and as such folic acid requirements increase during pregnancy. In fact, in a study published in 1991 in the prestigious medical journal Lancet, it was found that women who were considered at high risk of developing a common type of birth defect – neural tube defect – lowered their risk by 72 percent by means of folic acid supplementation. However, if a woman has the MTHFR defect, supplementation with regular folic acid may have little or no effect.

A doctor can order a simple lab test to determine if you have the MTHFR defect. Although there is no known way to make the activating enzyme work better, a nature-identical folate is available in supplement form, such as featured in the formula Methylator Plus 3.0. This ingredient, which goes by the acronym L-5-MTHF, works by bypassing the genetic defect and provides the body with the type of folic acid it needs.



The medical community is actively pursuing studies on MTHFR functioning. The following are a sample of the research studies on folic acid that have been published during the first half of 2011:

- Overtraining.** Research from Brazil has suggested a possible connection between poor folic acid metabolism and overtraining through an elevation in CRP, a marker for inflammation. (1)
- Cleft Palate.** A study in China showed a greater likelihood of cleft palate in those with the MTHFR defect. (2)
- Colitis.** Research from the Han province of China showed that patients with colitis were more likely to have the MTHFR defect. (3)
- DNA Repair Capacity and Oxidative Stress.** Italian researchers examined the effects of metals on DNA damage, reactive oxygen species, and the ability of DNA to repair. These markers were compared against various genes, including those with the MTHFR defect. They concluded that cellular damage was increased and DNA repair capacity was decreased in those with the MTHFR polymorphism. (4)
- Diabetic Neuropathy.** This study conducted in Iran observed the progression of diabetic neuropathy and the presence of both the MTHFR defect and another defect in endothelial nitric oxide synthase known as G894T. The two genes in combination were shown to contribute to an increased progression of diabetic neuropathy in patients with type II diabetes. (5)
- Antioxidant Effectiveness.** In an interesting study on runners and antioxidant consumption, Brazilian researchers examined the possibility that various genetic polymorphisms could impact the effectiveness of the

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antioxidants to alter lipid markers, inflammation, arterial stiffness and oxidative damage. They discovered that certain genetic predispositions, including the MTHFR defect, impaired the function of the antioxidant formula to exert its full effect. (6)

**Pregnancy Complications.** Researchers in Tunisia sought to determine if the MTHFR defect was involved in various pregnancy complications, including recurrent pregnancy loss, preeclampsia, placental abruption and intrauterine growth retardation. The researchers stated in the abstract, "We identified the carriage of the MTHFR A1298C polymorphism as a significant risk factor for vascular-related pregnancy complications." (7)

**Leukemia.** Scientists at the Texas Children's Cancer Center at Baylor University recently conducted a meta-analysis of 21 publications to better understand the connection between acute lymphoblastic leukemia (ALL) in children and the MTHFR polymorphism. Their data, covering more than 12,000 subjects, confirmed that the MTHFR gene should be included as a potential marker for ALL due to the significant correlation. (8)

**Colon Cancer.** Researchers at the Department of Clinical Oncology from Hospital Santa Rita in Brazil examined various polymorphisms for folate and methionine metabolism. They showed that all of these genetic variants, including the MTHFR gene, increased the likelihood of colon and rectal tumors. (9)

**Squamous Cell Carcinoma.** This study from Brazil used more than 800 subjects and examined the connection between the MTHFR defect and head and neck squamous cell carcinoma. Their observations stated that "MTHFR polymorphisms may contribute for increase risk for head and neck carcinoma." (10)

**Kidney Cancer.** Iranian researchers examined more than 450 subjects, 152 of whom had clear cell renal cell carcinoma (CCRCC). They showed that those with the MTHFR gene were at a significantly increased risk of this type of cancer – as much as 4.4 times higher risk. (11)

**Metabolic Syndrome.** In Greece, researchers examined 30 subjects with metabolic syndrome and 60 control subjects and concluded that MTHFR should be included as a risk factor. (12)

Check out this article for additional information on how to properly supplement your diet with the activated form of folic acid.

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**Re: The MTHFR Defect – Are You at Risk?**

Hey Coach! Lets say that you're thinking to yourself out loud. If you were a pregnant woman and you know you have the MTHFR gene, would you supplement with Methylator Plus 3.0 in pregnancy? Would it prevent the child from having the gene?

By **Guillermo Marranzini** on 6/8/2011 10:44 AM

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