MTHFR Genetic Defect

Do you have a genetic defect in the **MTHFR** gene?
Perhaps you have a family history of heart attack or stroke... Or you personally suffer from chronic migraine headaches, irritable bowel syndrome or depression… These are just some of the conditions that can be associated with a faulty enzyme called MTHFR.

What is MTHFR?
MTHFR stands for methyl-tetrahydrofolate reductase. It is an enzyme that is responsible for the **methylation** process throughout the body. If you have a genetic variant, it can cause this key enzyme to lose function. As a result, this can lead to medical problems. Currently, there are more than 50 recognized MTHFR variants. The two primary variants are called C677T and A1298.

What is Methylation?
Methylation is a process that occurs in all cells to convert one substance to another. When people with genetic MTHFR mutations are exposed to toxins, they have a more difficult time clearing the toxins from the body. If the toxins are not cleared, this may lead to illness. The process of proper methylation is responsible for:

- Activating/deactivating cellular activity
- Activating/deactivating enzymes
- Activating/deactivating neurotransmitters
- Turning on tissue repair
- Decreasing inflammation
- Activating/deactivating the stress response
- Reducing the aging process by protecting telomeres
- Detoxifying chemicals
- Facilitating production of glutathione which is the body's most important anti-oxidant
- Improving energy cycle by creating carnitine, CoQ10, ATP and supporting mitochondrial energy

What are common problems that can occur?
Certain mutations of MTHFR are commonly associated with specific health problems. Either of the two primary mutations can cause a wide variety of health problems.

- 677T variant is more commonly associated with cardiovascular problems, such as early heart disease and stroke
- 1298C variant is more commonly associated with a variety chronic illness

How are the genetic defects reported?

MTHFR anomalies are reported as **heterozygous** or **homozygous.**

- **Heterozygous** means you have one affected gene and one normal gene. This means that you inherited it from one parent.
  - It is estimated that your enzyme activity will be 40% less efficient than normal.
- **Homozygous** means 2 abnormal genes. You inherited one of the genes from each parent.
  - It is estimated that your enzyme activity will be 80-90% less efficient than normal. This can lead to serious consequences.
- **Compound Heterozygous.** In other words, you inherited one of each of the defects from each parent. This is the most serious combination: 677T/1298C. You are heterozygous to both anomalies.
  - Many chronic illnesses are linked to this anomaly including blood clots, irritable bowel syndrome, fibromyalgia, migraines, chemical sensitivity, and frequent miscarriage.

Other studies support the following possible loss of methylation functionality:

MTHFR C677T Heterozygous = 40% loss of function *
MTHFR C677T Homozygous = 70% loss of function *
MTHFR A1298C Heterozygous = 20% loss of function
MTHFR A1298C Homozygous = 40% loss of function **
MTHFR C677T & MTHFR A1298C heterozygous = compound heterozygous = 50% loss of function

Health Problems that may be related to MTHFR

The MTHFR gene mutation can make you susceptible to illness by lowering your body's ability to make glutathione. Glutathione is the body's primary antioxidant and detoxifier. People with MTHFR defects often have low glutathione, which makes them more susceptible to stress and less tolerant

to toxic exposures. An accumulation of toxins in the body increases stress and may lead to premature aging.

Conditions that may be associated with MTHFR gene mutations (Alphabetized)

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

You may not need medicine to treat a MTHFR defect!

Natural Treatments for MTHFR Mutation Symptoms

1. Consume More Natural Folate, Vitamin B6 and Vitamin B12

Consuming more natural folate (**not folic acid**, which is *synthetic* vitamin B9) can help with methylation. Methylated folic acid is the active form.

- Beans and lentils
- Leafy green vegetables like raw spinach
- Asparagus
- Romaine
- Broccoli (raw or lightly steamed)
- Avocado
- Bright-colored fruits, such as oranges and mangoes

Those with a MTHFR mutation are also more likely to be low in related vitamins, including **vitamin B6** and **vitamin B12**. Eat foods high in B vitamins, including quality protein foods, organ meats, nuts, beans, nutritional yeast and raw/fermented dairy products.

2. Treat Digestive Problems, Including Leaky Gut and Irritable Bowel Syndrome (IBS)

To improve digestive/gut health, the following dietary adjustments may be beneficial:

- Reduce intake of inflammatory foods, such as gluten, added sugar, preservatives, synthetic chemicals, processed meats, conventional dairy, refined vegetable oils, trans fats and processed/enriched grains (which often include synthetic folic acid).
- Increase intake of probiotic foods, which are fermented and supply "good bacteria" that aids in digestion.
- Consume other gut-friendly foods, including bone broth, organic vegetables and fruits, flaxseeds and chia seeds, and fresh vegetable juices.

Focus on consuming healthy fats only, like coconut oil or milk, olive oil, grass-fed meat, wild-caught fish, nuts, seeds, and avocado.

3. Reduce Anxiety and Depression

MTHFR mutations can negatively affect levels of neurotransmitters and hormones like serotonin, testosterone and estrogen, and are tied to higher incidences of mental disorders, including anxiety, depression, bipolar disorder, schizophrenia and chronic fatigue. High levels of stress may also make MTHFR mutation symptoms worse.

Tips for dealing with these conditions include:

- Supplementing with omega-3 fatty acids
    - Help to reduce inflammation
    - Beneficial for cognitive health
- Regularly practicing natural stress relievers
    - Include meditation, journaling, spending time outside, giving back or volunteering, praying, etc.
- Regularly exercising

    - Helps to improve hormonal balance and sleep quality
- Using soothing **essential oils**
    - Lavender, chamomile, geranium, clary sage and rose.
- Recreational drugs and alcohol, which can both make symptoms worse by interfering with methylation, should be eliminated.

4. Protect Heart Health

Studies show that homocysteine levels tend to rise with age, smoking and use of certain drugs, so the first step is to focus on taking care of yourself as you get older and eliminating the use of harmful substances. Additional tips for keeping your heart healthy include:

- Eating a healthy diet, especially one with plenty of high-fiber foods
- Getting regular exercise and keeping your weight in a healthy range
- Managing stress to prevent worsened inflammation
- Consider taking the following supplements to help improve blood flow, cholesterol and blood pressure: magnesium, omega-3s, CoQ10, carotenoids and other antioxidants, selenium, and vitamins C, D and E.

Glutathione is poorly absorbed so either the liposomal form or a precursor, called n-acetylcysteine (NAC) may be used. The choice of supplements will vary from patient to patient and should be done with professional supervision. Patients with these genetic mutations may be very sensitive to certain supplements, such as methyl-folate and methyl B12 or gluten, liposomal or acetyl-glutathione, which is the end product of the pathway.

There are prescription medicines, that also contain methyl-folate: Deplin, MetanX, CerefolinNAC are a few. Methyl B12 can also be given as shots, nasal sprays, and sublingually. If you suspect you have a genetic abnormality, contact your provider.