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MTHFR Genetic Testing

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MTHFR stands for "methylenetetrahydrofolate reductase," which is definitely a mouthful. MTHFR is an enzyme that helps process vitamin B9, which is also called folic acid or folate. Folate helps the body breakdown, utilize, and produce new proteins. A change or mutation in the MTHFR gene can cause the MTHFR enzyme to not work properly and therefore will result in a reduction in certain proteins. It is estimated that 55% of the population has one or more of the MTHFR genetic variants that may impair their ability to process folate. These mutations can have significant health implications.

One very important function of the methylenetetrahydrofolate reductase enzyme is in the changing of a chemical in the blood called homocysteine into another amino acid (a building block of protein) called methionine. Approximately 40 mutations in the MTHFR gene have been associated with hyperhomocysteinemia (a buildup in the blood of homocysteine). Elevated levels of homocysteine may be associated with cardiovascular disease which includes the hardening and narrowing of the arteries (atherosclerosis) and an increase in heart attacks, stroke, blood clots and possibly even Alzheimer's disease.

In addition to increasing risk for heart disease, elevated homocysteine levels may be related to renal disease, low thyroid hormone, psoriasis, low B vitamins (including B12 & B6), folic acid and effects on certain medications. This means that patients with elevated homocysteine levels or individuals with one or more of the MTHFR gene mutations should increase their B vitamins by eating more fruits, vegetables (especially green leafy vegetables), and grains which include folic acid. These individuals may also want to consider taking vitamin B supplements. In addition to having your homocysteine levels tested, MTHFR genetic testing may provide a more definitive risk assessment as well as specific treatment options from your physician.

It is worth noting that not all MTHFR mutations will have elevated homocysteine levels. Other conditions, symptoms, diseases or syndromes that may result from an inability to process folate and may be associated the MTHFR gene mutation include: cardiovascular disease, stroke, peripheral neuropathy, depression, OCD, bipolar disorder, schizophrenia and other mental disorders, anxiety, IBS, still births, pre-eclampsia, neural tube defects/cleft lip, fibromyalgia, chronic fatigue, dementia, migraines, schizophrenia, Parkinson's, miscarriage, infertility, cancer, autism or spectrum disorders and many other health conditions or diseases.

Our physicians at NaturoMedica are proactively involved with each patient in their individual and family risk assessment which may include advanced cardiovascular testing (including homocysteine levels), advanced nutrient testing, as well as the testing of the MTHFR genetic variants. At NaturoMedica, we look for the two most common types of MTHFR genetic mutations which are A1298C and C677T through a simple non-fasting blood draw which only needs to be ordered once in a lifetime.

While eating healthy and increasing various types of B vitamins in your diet may improve overall health, it is important to determine which particular B vitamins and other nutrients may be associated with the MTHFR genetic variants and which dosage(s) are optimal for each individual patient. Please feel free to consult with one of our physicians if you have questions or would like to have the genetic testing done for you or a family member.

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