What is MTHFR?

MTHFR (also called methylenetetrahydrofolate reductase) is an enzyme in your body that helps make the active form of folate. Folate is a form of Vitamin B which is also known as folic acid or Vitamin B9. Your body cannot make folate on its own, so it requires enzymes including MTHFR to make it from the foods that you eat.

MTHFR also has an important role in making amino acids, the building blocks of proteins, by helping convert the chemical homocysteine to methionine. Homocysteine is a harmful chemical made by your body when you eat meat that can damage the walls of your blood vessels and may affect your blood clotting. In contrast, methionine is an amino acid required by your body for normal functioning.

Doctors now know that some people have genetic differences, called mutations, in the gene for MTHFR that change the enzyme so that it does not work as well. If you have a mutation in MTHFR, you may not make as much folate as someone with a normal enzyme. Your body’s ability to convert homocysteine to methionine may also be affected. People with high homocysteine levels may be at higher risk of developing certain chronic diseases or may respond differently to some medications. Therefore, knowing if you have a mutation that weakens your MTHFR enzyme is important for your health.

How does knowing your MTHFR genotype help you and your doctor?

Your doctor can order a MTHFR genotype test to tell whether you have a normal or weak version of the MTHFR enzyme. The test looks at two places on the gene, called position 677 and position 1298, to see what your DNA sequence is at those places. Your sequence at position 677 (677 genotype) may be CC, CT or TT. Your sequence at position 1298 (1298 genotype) may be AA, AC, or CC. If you have the 677 TT genotype, you are at increased risk for elevated homocysteine levels and coronary artery disease.

If your doctor finds your MTHFR genotype forms a weaker version of the enzyme, specific dietary recommendations can be made to promote health and prevent harm. For example, people with the 677 TT genotype may need to include more sources of B vitamins in their diet, use other supplements to strengthen the methylation process, and/or have homocysteine levels monitored to be sure they aren’t too high. Knowing your MTHFR genotype can also help your doctor avoid recommending medications known to have worse side effects in people with a weak MTHFR enzyme.

When should you be tested for your MTHFR genotype?

Your doctor may test your MTHFR genotype if you have high homocysteine levels, if you have a family member who has a MTHFR mutation or if a family member developed blood clots or heart disease at a young age. Also, a MTHFR genotype test may be run before your doctor prescribes certain medications which affect how your body uses folate.

Can you do anything to change your MTHFR genotype?

No, unlike your blood pressure or cholesterol levels, there is nothing you can do to change your MTHFR genotype. However, knowing your MTHFR genotype will allow your doctor to make specific diet or treatment recommendations for you to promote your health.

How should you prepare for the MTHFR genotype test?

The MTHFR test does not require any special preparation. You do not need to fast prior to sample collection and you can take your medicines as you normally do.