MTHFR testing is no longer recommended for ANY reason.

Why not? Many people have one or two mutations in the MTHFR gene. However, having a mutation in the MTHFR gene does NOT change your medical care.

What is the MTHFR gene?
MTHFR is the gene responsible for making an enzyme in our cells, called methylenetetrahydrofolate reductase. This enzyme works with one of the B-vitamins, called folate. Together, MTHFR and folate help to reduce the level of homocysteine (a potentially harmful amino acid). Too much homocysteine in the body may be one risk factor for blood clots and heart disease. But MTHFR is not the only thing that determines homocysteine levels in the body. There are many other factors, both genetic and environmental, that determine how much homocysteine you have, and your risk for blood clots and heart disease.

I've been told I have an MTHFR mutation. What does that mean?
Everyone has two copies of the MTHFR gene, one copy from each parent. If you've been told you have an MTHFR mutation, it means you have a DNA change in either one or both copies of the MTHFR gene. The two most common MTHFR mutations are C677T and A1298C. However, there are other mutations that have also been found. The medical term used when you have one MTHFR mutation is heterozygous. Different terms are used when you have two mutations. When both copies of the gene have the same mutation, it is called homozygous. If there are two mutations that are not the same it is called compound heterozygous.

Someone in my family has an MTHFR mutation. Should I have testing?
There is no need to test for MTHFR status. Mutations in the MTHFR gene are very common, and finding one or two does not change your medical care. In 2013, the American College of Medical Genetics (ACMG) and The American Congress of Obstetrics and Gynecology (ACOG) published practice guidelines advising against MTHFR testing.

How common are MTHFR mutations?
MTHFR mutations are found in people worldwide. The chance to have a mutation in at least one copy of the gene is as high as 40% in some ethnic groups. Between 10%-15% of the Caucasian population and more than 25% of the Latino population have mutations in both copies of the MTHFR gene. One study on pregnant Chinese women found homozygous mutations in almost 30% of the women.
What happens when an MTHFR mutation is present?
The common MTHFR mutations cause a minor change in the DNA code. This makes the MTHFR enzyme work a little slower than usual. If MTHFR works too slowly, homocysteine might start to build-up in the body. Homocysteine build-up is slightly more likely to happen when a person has two copies of the C677T mutation. However, MTHFR mutations only have a small impact on your total homocysteine level. Most people with mutations in the MTHFR gene have normal levels of homocysteine.

What type of medical problems can be related to MTHFR mutations?
The relationship between the common MTHFR mutations and many medical problems has been wondered about for decades. There are some studies that suggest a possible increase in the risk for common medical problems. These include clotting problems, heart disease, stroke, high blood pressure, pregnancy complications, psychiatric disorders, autism, and certain types of cancer. These are all common conditions, which affect people with and without MTHFR mutations. Simply having an MTHFR mutation is not enough to determine your risk for any of these problems. Right now it is unknown what role, if any, the MTHFR gene mutations play in these conditions.

Does taking extra folate help a person with an MTHFR mutation?
Folate is a vitamin our body needs, but there is no known benefit for a person with an MTHFR mutation to take more than the recommended daily amount. It is important for everyone to get enough folate in their diet, regardless of their MTHFR status. It is especially important to get enough folate during pregnancy.

More about FOLATE and FOLATE SUPPLEMENTS
Folate is also called vitamin B9. It is found in the food you eat. Some foods high in folate include dark green vegetables (eg, spinach, broccoli, asparagus), legumes (lentils, beans, peas), and citrus fruits.

Folate supplements are the synthetic (man-made) forms of folate. The most common folate supplement is folic acid but there are also folic acid derivatives. These are a form of folic acid with slight chemical differences to make them easier for the body to absorb. If you do not get enough folate in the foods you eat, taking folic acid or a folic acid derivative makes sure your body has enough folate. Many breads and cereals have been fortified with folic acid. Prenatal vitamins usually include extra folic acid compared with regular vitamins.

Reference: 2013 ACMG/ACOG Practice Guidelines
The information is not intended to diagnose health problems or to take the place of professional medical care. If you have persistent health problems or if you have further questions, please consult your health care provider.

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