Pediatric Division of Genetics and Metabolism

The Division of Pediatric Genetics and Metabolism at UNC does **NOT** offer *MTHFR* testing and evaluation. For additional information and recommendations about *MTHFR,* please see below.

What is methylene tetrahydrofolate reductase (*MTHFR*)?

MTHFR is a gene, not a disease or genetic condition. The *MTHFR* gene serves many functions. One of its most important functions is providing instructions to create an enzyme called methylentetrahydrofolate reductase. This enzyme plays an important role in the metabolism (chemical reactions within the body) of folic acid (also called vitamin B9), meaning it helps the body process this essential nutrient.

Can having changes in the *MTHFR* gene cause problems or change medical care?

Some changes in the *MTHFR* gene reduces or will completely turn off the enzyme. This leads to elevated levels of a specific amino acid called homocysteine.

We also know that there are two common naturally occurring changes in the *MTHFR* gene, called C677T and A1298C. These are called polymorphisms as they do NOT necessarily affect the function of the *MTHFR* gene and do NOT directly cause medical issues or symptoms. It is important to understand that these two polymorphisms are NOT associated with a metabolic condition called homocystinuria, which causes problems with the eyes, brain, and other portions of the body.

There has been a lot of research about the potential health impacts of having these common *MTHFR* changes and the findings remain conflicting and inconclusive. The existing scientific data does NOT support the claims that common *MTHFR* changes impact human health. At this time, there are currently no changes in medical care that are offered or recommended to individuals with the C667T and A1298C common *MTHFR* changes.

How common are the C667T and A1298C changes?

It is very common within the general population to carry either one or two of these changes in the *MTHFR* gene. In some ethnicities, about 20-40% of individuals are carriers for just one of these common changes and about 10-20% have two copies of these common changes.

Do individuals need to be tested for the common *MTHFR* changes?

Established medical guidelines do NOT recommend *MTHFR* testing because these gene changes are so common in the general population and do NOT directly cause medical issues or impact clinical care.

What about *MTHFR* in pregnancy?

Standard practice recommends that all women of childbearing age take a multivitamin containing folic acid prior to conception. Folic acid supplementation reduces the chance of neural tube defects (when the spinal column fails close properly in early pregnancy). Having or not having the common *MTHFR* changes do NOT alter recommendations for folic acid supplementation.

What is all the hype about *MTHFR* online?

Websites and blogs are filled with misinformation about *MTHFR*. Some websites and alternative health practitioners have made bold claims that common changes in *MTHFR* gene can cause many health conditions, ranging from blood clots and cancer to autism and migraines. However, little to no peer-reviewed scientific research supports these claims.

Where can I find more information about *MTHFR*?

For more information, please refer to the American College of Medical Genetics and Genomics practice guidelines for *MTHFR.* Additional professional guidelines can be found through the American Heart Association and American Congress of Obstetricians and Gynecologists. Links to these guidelines can be found on our website.

Helpful Links:

American College of Medical Genetics and Genomics
https://www.acmg.net/docs/MTHFR_gim2012165a_Feb2013.pdf

American Heart Association
http://circ.ahajournals.org/content/111/19/e289

American Congress of Obstetricians and Gynecologists
https://www.acog.org/clinical/clinical-guidance/practice-bulletin/articles/2018/07/inherited-thrombophilias-in-pregnancy