VCU Health™

*Department of Pathology Laboratories*

Notification Date: July 15th, 2025
Effective Date: July 9th, 2025

# MTHFR (Methylene-tetrahydrofolate Reductase)– Test Discontinued

**Orderable:** MTHFR [LAB1231492]

**After clinical and literature review by the VCU Health Laboratory Utilization Committee, effective July 9, 2025, MTHFR genotyping can no longer be ordered through VCU Health Pathology.**

## Explanation:

This decision has been made after a review of the literature and our own clinical experience here at VCU Health.

There are two common variants in MTHFR: C677T and A1298C. These are known to decrease activity of the MTHFR enzyme. It is common in the general population to have these variants on one, or both, copies of the MTHFR gene. There are no health risks known to be associated with having one copy of either the C677T and A1298C variant (heterozygous), two copies of the A1298C variant (homozygous), or having one copy of each variant (compound heterozygous). Health problems can occur if a person is homozygous for the C677T variant and has elevated fasting plasma total homocysteine levels. These health concerns include mild to moderate risk of blood clots, stroke, miscarriage, and neural tube defects in offspring. Elevated homocysteine levels should be managed accordingly, regardless of MTHFR genotype.

The American College of Medical Genetics and Genomics recommends against MTHFR polymorphism genotyping for the clinical evaluation of thrombophilia and recurrent pregnancy loss, as well as for at-risk family members. Additionally, MTHFR status does not change the recommendation that women of childbearing age should take the standard dose of folic acid supplementation to reduce the chance of neural tube defects. The ACMG Practice Guidelines are available for your review.

https://www.nature.com/articles/gim2012165

**For questions, please contact Pathology Client Services at 804-828-7284 (8-PATH).**