Indiana University Health Molecular Pathology Laboratory

MTHFR

CERNER ORDERABLE
MTHFR Mutation PCR

CPT CODE
81291

CLINICAL UTILITY
Human methylenetetrahydrofolate reductase (MTHFR) is a key enzyme in the metabolism of homocysteine. High levels of homocysteine in the blood (hypercysteinemia) is a risk factor for deep vein thrombosis and cerebrovascular disease; including arterial thrombosis, atherosclerosis, and coronary heart disease, and has also been reported in neural tube defects, and folate metabolism disorders.
MTHFR deficiency is caused by a point mutation within the MTHFR gene. IU Health Molecular Pathology lab tests for the most common mutations: C677T and A1298C[1].

METHODOLOGY
PCR/ Melting Curve Analysis

SPECIMENS
Peripheral blood drawn in a 3 mL or 6 mL EDTA tube. Minimum acceptable volume is 2 mL. For pediatric patients, an EDTA micro-container may be used. Minimum acceptable pediatric volume is 500 µL. Do not spin.

SPECIMEN STABILITY and SHIPPING
Store and ship whole blood specimen refrigerated. Do not spin.

CAUSES FOR REJECTION
Clotted or contaminated sample; if collected in any other anticoagulant.

SPECIFICITY
MTHFR C677T and A1298C mutations are the only mutations identified in this assay.

ASSAY RANGE
Genotype differentiation between wildtype (normal), heterozygous, or homozygous mutation for C677T and A1298C

TURNAROUND TIME
Thursday, 7 days

1. Reference information can be found in the Indiana University Health Molecular Assay Procedures.