MTHFR - Thermolabile Variant, DNA Analysis

Order Name: **MTHFR**
Test Number: 1515625
Revision Date: 10/22/2024

TEST NAME	METHODOLOGY	LOINC CODE
MTHFR Interpretation	Interpretive information.	

SPECIMEN REQUIREMENTS

Specimen	Specimen Volume (min)	Specimen Type	Specimen Container	Transport Environment
Preferred	**7mL (3mL)**	**Whole Blood**	**EDTA (Lavender Top)**	**Room Temperature**
Alternate 1	**7mL (3mL)**	**Whole Blood**	**ACD Solution A or B (Yellow Top)**	**Room Temperature**
Alternate 2	**2 Swabs**	**Buccal swab**	**Labcorp buccal swab kit**	**Room Temperature**

Instructions

Specimen Requirements

Specimen: Whole blood or LabCorp buccal swab kit (buccal swab collection kit contains instructions for use of a buccal swab)

Volume: 7 mL whole blood or LabCorp buccal swab kit (Minimum Volume 3 mL whole blood or two buccal swabs)

Container: Lavender-top (EDTA) tube, yellow-top (ACD) tube, or LabCorp buccal swab kit

Storage Instructions: Maintain specimen at room temperature or refrigerate.

Causes for Rejection: Frozen specimen; hemolysis; quantity not sufficient for analysis; improper container; one buccal swab; wet buccal swab.

GENERAL INFORMATION

Expected TAT

6 - 10 days

Clinical Use

Methylenetetrahydrofolate Reductase (MTHFR) Thermolabile Variant, DNA Analysis
Variants Analyzed: c.665C>T (p. Ala222Val), legacy name: C677T; and c.1286A>C (p. Glu429Ala), legacy name: A1298C. Results must be combined with clinical information for the most accurate interpretation. Molecular-based testing is highly accurate, but as in any laboratory test, diagnostic errors may occur. False positive or false negative results may occur for reasons that include genetic variants, blood transfusions, bone marrow transplantation, somatic or tissue-specific mosaicism, mislabeled samples, or erroneous representation of family relationships. This test was developed and its performance characteristics determined by Labcorp. It has not been cleared or approved by the Food and Drug Administration.

Notes

Hyperhomocysteinemia is multifactorial, involving genetic, clinical, and environmental risk factors. Reduced enzyme activity of methylenetetrahydrofolate reductase (MTHFR) is a genetic risk factor for hyperhomocysteinemia, particularly when serum folate levels are low. There are two common variants in the MTHFR gene that can decrease enzyme activity: c.665C>T (p. Ala222Val), legacy name C677T, and c.1286A>C (p. Glu429Ala), legacy name A1298C. These variants do not independently increase risk of conditions related to hyperhomocysteinemia in the absence of elevated homocysteine levels. Measurement of total plasma homocysteine is recommended. Patients should share their MTHFR genotype with physicians who are making decisions regarding chemotherapy treatments that depend on folate, such as methotrexate. Guidelines do not recommend genotyping of these two MTHFR variants in the evaluation of venous thromboembolism or obstetric risk due to limited evidence of clinical utility. Genetic coordinators are available for health care providers to discuss results and for information on how to order additional testing, if desired, at 1-800-345-GENE.

CPT Code(s)

81291

Service Provided By

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