LabCorp
Laboratory Corporation of America

Specimen Number	Patient ID	Control Number	Account Number	Account Phone Number	Rte

Patient Last Name			
Patient First Name	Patient Middle Name		
Patient SS#	Patient Phone	Total Volume	
Age (Y/M/D)	Date of Birth / Sex	Fasting	

Patient Address	Additional Information

Date and Time Collected	Date Entered	Date and Time Reported	Physician Name	NPI	Physician ID

Tests Ordered

General Comments

TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB

MTHFR

MTHFR, DNA Analysis 01

Result: C677T/C677T

Two copies of the same mutation (C677T and C677T) identified

Interpretation:
This patient's sample was analyzed for the MTHFR mutations C677T and A1298C. Two copies of the C677T mutation were identified. Results for the A1298C mutation were negative. The diagnosis of hyperhomocysteinemia can not rely on DNA testing alone but should take into consideration clinical findings and other studies, such as serum homocysteine levels. Because MTHFR mutations and their associated risks are inherited, genetic counseling and testing of at-risk family members should be considered.

FINAL REPORT

Sample test results. Actual results may vary.

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Additional Information:					01

Genetic counselors are available to discuss these results with health care providers at 1-800-345-GENE

The MTHFR enzyme is responsible for creating the circulating form of folate. Folate is important in homocysteine regulation. Defects in the MTHFR enzyme can indirectly cause elevated homocysteine levels. The C677T mutation in the MTHFR gene can cause elevated homocysteine levels in individuals with insufficient folate, particularly when there are two mutations present. The A1298C mutation has not been associated with elevated homocysteine levels unless a C677T mutation is also present. Elevated serum homocysteine levels have been associated with an increased risk of cerebrovascular disease, coronary artery disease, myocardial infarction, and venous thrombosis. In women, pregnancy complications and an increased risk of fetal open neural tube defects have also been reported. The relationship between these conditions and MTHFR mutations is controversial.

Dietary folic acid, B6 and B12 supplementation has been suggested to lower homocysteine levels in some people. Folic acid supplementation has been shown to reduce the recurrence of neural tube defects.

Methodology:

DNA analysis of the MTHFR gene was performed by PCR amplification followed by restriction analysis. The diagnostic sensitivity is >99% for both. Molecular-based testing is highly accurate, but as in any laboratory test, rare diagnostic errors may occur. All test results must be combined with clinical information for the most accurate interpretation.

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References: 01

Eskes TKAB. (1998). Nutrition Review 56:236-244.
Rozen R. (1997). Thromb Haemost 78:523-526.
Botto and Yang. (2000). Am J Epidemiol 151:862-877.

Kenneth J. Friedman, Ph.D.
Lauren Kam-Morgan, Ph.D.
Li Cai, Ph.D.
Marcia Eisenberg, Ph.D.
Suzette M. Huguenin, Ph.D.
Dorothy M. Adcock, M.D.
Val. V. Zvereff, Ph.D.

FINAL REPORT