FOUNDATION LABORATORY

ANNOUNCEMENT

Date: May 18, 2012

Dear Valued Clients:

Foundation Laboratory is pleased to announce that effective May 17, 2012 serum MTHFR will be performed in-house. This FDA approved assay is performed using multiplex PCR with solid phase electrochemical methodology on GenMark Dx eSensor platform and detects 577C>T and 1298A>C mutations. This assay is recommended by The American College of Medical Genetics (ACMG).

MTFHR, 5, 10-methylenetetrahydrofolate, is used to convert homocysteine (a potentially toxic amino acid) to methionine by enzyme methionine synthase. The enzyme is coded by gene with the symbol MTHFR on chromosome 1 location p36.3 in humans. The number of polymorphisms are up to 24 and two of the most investigated are C677T (rs1801133) and A1298C (rs1801131) single nucleotide polymorphisms (SNP).

Approximately 50% of patients with hereditary thrombophilia demonstrate thrombotic event in presence of additional risk factors such as immobilization, surgery, or orthopedic trauma. Because folate is vital for conversion of homocysteine to methionine, patients with severe MTHFR deficiency can develop homocysteinemia which is a risk factor for cardiovascular disease (CAD). MTHFR DNA testing is indicated for prediction of hyperhomocysteinemia , patients with history of venous thrombosis, CAD and/or stroke, to determine the genetic cause of early onset of arterosclerotic vascular disease, identification of individuals at high risk for methotrexate sensitivity due to prolonged use of methotrexate and optimization of their therapy, history of pregnancy complications including neural tube defect, still births, and/or recurrent pregnancy loss, or relatives of individuals with hyperhomocysteinemia or MTHFR gene mutation.

MTHFR C677T heterozygote frequency is up to 40% in Caucasian population. Homozygote frequency is 5-10% in Caucasian population, and 1.4% in African American population. C677T homozygotes have mildly elevated homocysteine levels and modest increase in risk of thrombosis and arteriosclerosis.

MTHFR A1298C frequency is as high as 30% in the general Caucasian population. In conjunction with C677T or other genetic thrombotic factors (e.g., Factor V Leiden), the risk for venous thrombosis is increased. Heterozygosity for 1298C alone does not result in hyperhomocysteinemia. A1298C/C677T heterozygote may equal or exceed C677T homozygote symptoms.

FOUNDATION LABORATORY

Specimen Requirements:

- 1 full whole blood lavender top tube, refrigerated.

Turn Around Time:

- 10 days

For supplies and other needs please contact your Foundation Laboratory representative.

Sincerely,

Reza M. Massoumi, Ph.D.
Director of Technical Operations