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ACCT. NO.EA34168647
COLLECT. 04/03/13 17:00
RECEIVED 04/03/13 23:05
REPORTED 04/04/13 14:48

PATIENT MARTIN,JOYCE
PT I.D.
SEX F AGE 57Y
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85932

Req#: 17261546

DOB 07/13/55

Test Procedure	Results	Reference Range
MTHFR 2 MUTATIONS		Lab No:100
C677T MUTATION	HETEROZYGOUS	SEE DETAIL
A1298C MUTATION	HETEROZYGOUS	SEE DETAIL

POSITIVE FOR COMPOUND HETEROZYGOTE MTHFR MUTATIONS C677T AND A1298C

One copy of the MTHFR gene mutation C677T was detected and one copy of the MTHFR gene mutation A1298C was detected. This genotype is predicted to be associated with increased plasma homocysteine levels and an increased risk for coronary heart disease and venous thrombosis as well as for toxicity from medications affecting folate metabolism.

Mutations in the MTHFR gene (C677T and A1298C) correlate with reduced enzyme activity; however, only homozygotes for C677T or compound heterozygotes for C677T/A1298C have significantly elevated plasma homocysteine levels and increased risk for premature cardiovascular disease. These individuals may also show toxicity from medications (i.e. methotrexate) that affects folate metabolism.

Appropriate use of this test requires medical interpretation and counseling within the overall clinical care of the individual patient.

Methodology: PCR and solid phase electrochemical detection (voltammetry).

** END OF PATIENT REPORT **