TEST ID: Methylenetetrahydrofolate Reductase (MTHFR)

DESCRIPTION: Genetic testing for MTHFR 677C>T and 1298A>C gene mutations

COMPONENTS
- MTHFR 677C>T mutation
- MTHFR 1298A>C mutation

CLINICAL USE

Mutation 677C>T: This SNP or mutation is most commonly associated with: cardiovascular disease (including heart attack, stroke and blood clots) as well as peripheral neuropathy, miscarriages, neural tube defects (spina bifida) and other congenital birth defects.

Mutation 1298A>C: This SNP or mutation is most commonly associated with: chronic disease, e.g. depression, fibromyalgia, chronic fatigue syndrome, migraines, IBS (Irritable Bowel Syndrome), dementia, OCD, bipolar, schizophrenia, and more.

SPECIMEN INFORMATION

COLLECTION
Whole blood in sodium citrate tube.

SPECIMEN STABILITY
Stable at room temperature for 1 week.

REJECTION CRITERIA
Blood sample is over 1 week old; grossly hemolyzed, icteric, and lipemic specimens; improper anticoagulant used.

METHOD
Real-time SSP-PCR (Polymerase Chain Reaction using Sequence Specific Primers) followed by melt curve analysis

RESULT INTERPRETATION

Presence or absence of MTHFR 677C>T and MTHFR 1298A>C mutations are reported. Homozygosity (both copies of the gene contain the mutation) or heterozygosity (only one of two copies of the gene contains the mutation) of each mutation is reported for the affected individuals. Risk factors and percent of enzyme activity associated with each combination of genotypes are reported.

CPT CODES

| MTHFR 677C>T | 81291 |
| MTHFR 1298A>C | 81291 |

REFERENCES


