

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

Homocysteine and MTHFR C677T polymorphism in children and adolescents with psychotic and mood disorders. Kevere L, Purvina S, Bauze D, Zeibarts M, Andrezina R, Piekuse L, Brekis E, Purvins I. *Nord J Psychiatry*. 2013 Apr 16

Hyperhomocysteinemia and of Methylenetetrahydrofolate Reductase (C677T) Genetic Polymorphism in Patients with Deep Vein Thrombosis. Brezovska-Kavrakova J, Krstevska M, Bosilkova G, Alabakovska S, Panov S, Orovchanec N. *Mater Sociomed*. 2013;25(3):170-4. doi: 10.5455/msm.2013.25.170-174.

Genetic polymorphisms and the risk of myocardial infarction in patients under 45 years of age. Sakowicz A, Fendler W, Lelonek M, Sakowicz B, Pietrucha T. *Biochem Genet*. 2013 Apr;51(3-4):230-42. doi: 10.1007/s10528-012-9558-5.

Lewis SJ, Lawlor DA, Davey Smith G, et al. (2006). "The thermolabile variant of MTHFR is associated with depression in the British Women's Heart and Health Study and a meta-analysis". *Mol. Psychiatry* 11 (4): 352-60. doi:10.1038/sj.mp.4001790. PMID 16402130.

Maternal and infant gene-folate interactions and the risk of neural tube defects. Etheredge AJ, Finnell RH, Carmichael SL, Lammer EJ, Zhu H, Mitchell LE, Shaw GM. *Am J Med Genet A*. 2012 Oct;158A(10):2439-46. doi: 10.1002/ajmg.a.35552.

Methylenetetrahydrofolate reductase polymorphism affects the change in homocysteine and folate concentrations resulting from low dose folic acid supplementation in women with unexplained recurrent miscarriages. Nelen WL, Blom HJ, Thomas CM, Steegers EA, Boers GH, Eskes TK. *J Nutr*. 1998 Aug;128(8):1336-41.

Schwahn B, Rozen R (2001). "Polymorphisms in the methylenetetrahydrofolate reductase gene: clinical consequences". *Am J Pharmacogenomics* 1 (3): 189-201. doi:10.2165/00129785-200101030-00004. PMID 12083967.