



GENOMIC_ASSESSMENTS

BLOOD - EDTA

Result Range Units

MTHFR Gene Mutation

MethyleneTetraHydroFolate Reductase (MTHFR) Gene Mutation.

MTHFR Gene Mutation (C677T): Heterozygous for the mutation.

MTHFR Gene Mutation (A1298C): Heterozygous for the mutation.

Comment:

The patient has one copy of the MTHFR C677T mutation and one copy of the A1298C mutation.

This is associated with increased in plasma homocysteine levels.

MethyleneTetraHydroFolate Reductase (MTHFR) is a regulatory enzyme in folate-dependent homocysteine remethylation.

A common polymorphism in the MTHFR gene at position 677 is associated with a thermolabile enzyme with decreased activity. The prevalence of the homozygous mutation ranges from 8- 18% in various populations.

Clinically, homozygotes for the mutation have an increased risk of thromboembolism as well as premature vascular disease.

A second mutation (A1298C) has also been described. This mutation is associated with an increased risk of thromboembolism, when only found together with the C677T mutation.