



JOE DOE

Date of Birth: 08-Jan-1985

Sex: M

Collected: 31/Oct/2019 Received: 31/Oct/2019 INTERNATIONAL PATIENT

1000

Lab id: **3640442** UR#:

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GENOMIC ASSESSMENTS

BLOOD - EDTA

Result

Unite

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.

Range

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.

Tests ordered: IMPEI,CFee,MTHFR