FACTOR V LEIDEN MUTATION ANALYSIS

FACTOR V LEIDEN MUTATION ANALYSIS (Real Time PCR) Factor V Leiden Mutation Prothrombin gene Mutation MTHFR gene Mutation

Interpretation

RESULT	COMMENTS
Homozygous mutation detected	Both copies of the gene carry mutation
Heterozygous mutation detected	One copy of the gene carries mutation
Not Detected	Mutation not detected

Note:

1. This assay detects mutation in 3 genes namely Factor V Leiden (R506Q), Factor II Prothrombin gene mutation (G20210A) & MTHFR (C677T).
2. This is an in-house developed assay
3. Test conducted on Whole blood
4. Genetic counseling available.

Comments

A mutational defect in factor V causes APC (Activated Protein C) resistance which can be homozygous or heterozygous. Factor V Leiden increases the relative risk of thrombosis by 2-3 fold in the heterozygous condition and by 8-10 fold in the homozygous individual. Factor V Leiden mutation is a risk factor for venous as well as arterial thrombosis.

Homozygous mutation for MTHFR (Methylenetetrahydrofolate reductase) is associated with hyperhomocysteinemia which is an independent risk factor for vascular disease. Indian studies suggest that heterozygosity for MTHFR C677T is also associated with elevated homocysteine levels. Multiple mutations have synergistic effects.

Uses

Used as a thrombosis risk factor in patients prior to major surgery, pregnancy, post partum, oral contraceptive use, estrogen replacement therapy, transient ischemic attacks, premature stroke, peripheral vascular disease, pulmonary embolism & family history of thrombosis or known Factor V mutation.