Patient ID : p\_id Name : p\_name Age/Sex : age\_sex
Ref. by : doctor\_ref Lab no : Date : test\_date
Thrombophille profile
Prothrombin (Factor II) Mutation Assay (Qualitative) ^
PCR and Gel Electrophoresis
Specimen type: EDTA P. Blood
RESULT :
Prothrombin Mutation 20210G>A IN RANGE OUT OF RANGE
No Mutation Detected \*\*\*\*\*\*
Result:
Prothrombin 20210G>A mutation was not detected in the leukocytes of the specimen
Interpretation:
Factor II (RefSeq NM\_000506) codes for Prothrombin, which is a vitamin K dependent proenzyme that functions in the blood coagulation cascade. Prothrombin G20210A mutation occurs in the noncoding region of the Factor II gene and is the second most common cause of inherited thrombophilia after FVL mutations. It results in elevated levels of plasma prothrombin leading to hypercoagulability. Heterozygous individuals have a 2-4 fold increase in thrombotic risk. The test may be used for evaluation of patients with early onset VTE, as a thrombosis risk factor in patients prior to major surgery, to determine the cause of recurrent second or third trimester pregnancy loss, screening for risk of thrombosis before Oral contraceptive use and estrogen replacement therapy.
Test Attributes and Limitations:
Samples must be received at the laboratory under appropriate conditions within 72hrs of aspiration to ensure preservation of high molecular weight DNA. PCR is a highly sensitive technique; reasons for apparently contradictory results may be due to improper quality control during sample collection, selection of inappropriate specimen and/or presence of PCR inhibitors.
MTHFR Mutation Assay (Qualitative) ^
PCR and Gel Electrophoresis
Specimen type: EDTA P. Blood
RESULT :
MTHFR Mutation 677C>T IN RANGE OUT OF RANGE
No Mutation Detected \*\*\*\*\*\*
Result:
MTHFR 677C>T mutation was not detected in the leukocytes of the specimen
Interpretation:
MTHFR gene (RefSeq NM\_005957) codes for the enzyme Methylenetetrahydrofolate reductase. A genetic polymorphism in MTHFR gene (677C>T) results in the formation of the enzyme that has reduced activity and causes decreased metabolism of Homocysteine. Eleveated plasma Homocysteine has been associated with Venous Thromboembolism (VTE) and atherosclerotic vascular disease. Testing for MTHFR mutation may be useful for determining genetic causes for early onset hyperhomocysteinemia and for predicting sensitivity to Methotrexate and antifolate medications. Homozygosity for 677C>T predicts increased risk for atherosclerotic vascular disease and VTE. Such patients are also at risk for Methotrexate intolerance and may require dosage adjustment.
Test Attributes and Limitations:
Samples must be received at the laboratory under appropriate conditions within 72hrs of aspiration to ensure preservation of high molecular weight DNA. PCR is a highly sensitive technique; reasons for apparently contradictory results may be due to improper quality control during sample collection, selection of inappropriate specimen and/or presence of PCR inhibitors.
Factor V Leiden Mutation Assay (Qualitative) ^
PCR and Gel Electrophoresis
Specimen type: EDTA P. Blood
Specimen type: EDTA P. Blood
RESULT :
Factor V Leiden Mutation 1691G>A IN RANGE OUT OF RANGE
No Mutation Detected \*\*\*\*\*\*
Result:
Factor V Leiden (1691G>A) mutation was not detected in the leukocytes of the specimen
Interpretation:
Factor V is a protein of the Coagulation system. It is coded by the gene FV (RefSeq NM\_000130). A mutational defect in Factor V (R506Q) causes APC (Activated Protein C) resistance which can be homozygous or heterozygous. Factor V Leiden increases the relative risk of thrombosis by 5-10 fold in the heterozygous condition and by 50-100 fold in the homozygous individual. The lifetime risk for DVT is 12-20% for Heterozygotes and 80% for Homozygotes. Factor V Leiden Mutation is a risk factor for venous as well as arterial thrombosis. It is the most common genetic risk factor for thrombosis and accounts for >90 percent of APC resistance. The Test may be used as a thrombosis risk factor in patients prior to major surgery, to determine the cause of recurrent second or third trimester pregnancy loss, screening for risk of thrombosis before Oral contraceptive use, estrogen replacement therapy and for presymptomatic evaluation of individuals with a family history of thrombosis or a family member identified to have FVL.
Test Attributes and Limitations:
Samples must be received at the laboratory under appropriate conditions within 72hrs of aspiration to ensure preservation of high molecular weight DNA. PCR is a highly sensitive technique; reasons for apparently contradictory results may be due to improper quality control during sample collection, selection of inappropriate specimen and/or presence of PCR inhibitors.
NOTE : This test was processed at third party lab.
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