



TEST: PROTHROMBIN GENE 20210 MUTATION (PGM)

PRINCIPLE:

Prothrombin is the gene which encodes serine protease thrombin, a key enzyme in the processes of hemostasis and thrombosis. Recently, a point mutation (G→A 20210) has been associated with deep vein thrombosis. Carriers of the mutation have a 2.8 fold increased risk for venous thrombosis.

SPECIMEN REQUIREMENTS:

10mL whole blood collected in lavender top EDTA tubes (two 5ml tubes). Specimen should be delivered to the laboratory within 72 hours at room temperature. Peripheral blood specimens that are clotted, have not been collected in EDTA, or frozen are not acceptable.

METHOD:

Polymerase Chain Reaction (PCR) and reverse hybridization.

REFERENCES:

1. Poort, S.R., Rosendaal, F.R., Reitsma, P.H., Bertina, R.M. 1996 A common genetic variation in the 3'-untranslated region of the prothrombin gene is associated with elevated prothrombin levels and an increase in venous thrombosis. *Blood* 88:3698-3703
1. Rosendaal, F.R., Doggen, C.L.M., Zivelin, a., Arruda, V.R., Aiach, M., Siscovick, D.S., Hillarp, A., Watzke, H.H., Bernardi, F., Cumming, A.M., preston, F.E., Retsma, P.H. 1998. Geographic distribution of 20210 G to A prothrombin variant. *Thromb.Haemost* 79:706-708
2. Rosendaal, F.R., Doggen, C.L.M., Zivelin, a., Arruda, V.R., Aiach, M., Siscovick, D.S., Hillarp, A., Watzke, H.H., Bernardi, F., Cumming, A.M., preston, F.E., Retsma, P.H. 1998. Geographic distribution of 20210 G to A prothrombin variant. *Thromb.Haemost* 79:706-708
3. Nguyen, A. 2000. Prothrombin G20210A polymorphism and thrombophilia. *Mayo Clin. Proc.* 75:595-604.
4. Marisa, B., Marques, M.D. Testing for genetic predisposition to venous thrombosis. *MLO*, January 2002

Normal Range: Reported as: Normal, Heterozygous Mutated or Homozygous Mutated

Turnaround time: 10 business days