

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. To detect the point mutation, a 345 bp fragment of the prothrombin gene surrounding nucleotide 20210 was amplified by PCR using mismatched primer introducing allele specific restriction enzyme site following by digestion with Hind III. The amplified fragments from the mutation allele yield 2 fragments of 322 bp and 23 bp. The results then can be visualized by gel electrophoresis

SPECIMEN REQUIREMENTS:

Collect two to three tubes of blood (lavender) with EDTA. Heparinized blood cannot be used. Specimen should be delivered to the laboratory immediately, within 24 hours. Store or send 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)

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 PGM Normal, GM Heterozygous or PGM Homozygous

PGM Normal	345 bp fragment
PGM Heterozygous	345, 322, 23* bp fragment
PGM Homozygous Mutated	322, 23* bp fragment

23* bp is not visible on gel.

Turnaround time: Two Weeks