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Prothrombin DNA Screen

Background

Prothrombin (factor II) is one of the blood coagulation factors. A variant (base 20210G->A) in the 3' untranslated region of the prothrombin gene (*F2*) is associated with an increased risk for venous thrombosis. Approximately 20% of Dutch patients with a family history of venous thrombosis are heterozygotes for the 20210A allele, as compared to about 1% of healthy controls. In a population-based study, the 20210A allele appears to increase the risk of venous thrombosis about 3-fold for adults of both sexes. This test determines the presence or absence of the 20210G (normal) and 20210A (variant) alleles in the prothrombin gene.

Indications for Testing

-Evaluate cause of recurrent or familial venous thrombosis
-Presymptomatic testing in family known to carry a 20210A variant allele
-Evaluate recurrent pregnancy loss

Ordering

1. Obtain blood sample - (see Sample Requirements below).

2. Fill out a Clinical Lab Request - Genetics for each patient.
   (Available at http://depts.washington.edu/labweb/Divisions/MolDiag/MolDiagGen/index.htm).
   - Request: "Prothrombin 20210A"

3. Call Laboratory Medicine Community Services at (206)598-6066 to arrange the best method of shipment.

Sample Requirements and Specimen Handling

Whole blood - EDTA (purple top) - 5 mL.
Samples should be received within 72 hours of collection.
Samples may be refrigerated until shipped.
For prenatal diagnosis specimens, consult laboratory.
Heparin (green top) tubes are not acceptable.

Test Frequency and Reporting

Test results usually within 1-2 weeks of specimen receipt.
A written interpretative report is provided.

Reference

Poort SR, Rosendaal FR, Reitsma PH, Bertina RM. A common genetic variation in the 3'-untranslated region of the prothrombin gene is associated with elevated plasma prothrombin levels and an increase in venous thrombosis. Blood 88:3698-3703, 1996.

McGlennen RC, Key NS. Clinical and laboratory management of the prothrombin G20210A mutation. Arch Pathol Lab Med 126:1319-1325, 2002.

Seligsohn U, Lubetsky A. Genetic susceptibility to venous thrombosis. N Engl J Med 344:1222-1229, 2001.

Rey E, Kahn SR, David M, Shrier I. Thrombophilic disorders and fetal loss: a meta-analysis. Lancet 361:901-908, 2003.