

FACTOR II (PROTHROMBIN)

DESCRIPTION

Mendelian Inheritance in Man number: [176930](#)

Factor II (also called prothrombin) is a protein that is required for the clotting process in the blood. The G to A mutation at nucleotide 20210 in the prothrombin gene causes an overproduction of this protein and can increase risk for venous thrombotic events (VTE). Heterozygous prothrombin mutations occur in about 2% of the US white population. The mutation is uncommon in African Americans, and is rare in other ethnic groups, such as Asians, Africans, and Native Americans. Individuals homozygous for prothrombin mutations are uncommon, with an expected occurrence of approximately 1 in 10,000. Clinical presentation varies among individuals, and is dependent on other factors, such as environmental influences. Having a heterozygous prothrombin mutation increases the risk of developing a first VTE by about two- to threefold. Having homozygous prothrombin mutations increases the risk further, but it is not yet known how much the risk is increased

INDICATIONS FOR DIRECT TESTING

- A first VTE before 50 years of age
- A first unprovoked VTE at any age
- Recurrent VTE
- Venous thrombosis at unusual sites (such as cerebral, mesenteric, portal, and hepatic veins)
- VTE during pregnancy, the puerperium, or in association with oral contraceptive use or hormone replacement therapy
- A first VTE and a strong family history of VTE
- At risk individuals in families with known prothrombin mutations

TESTING METHODOLOGY

The Prothrombin molecular genetic test is an allele-specific based approach described by [Hezard *et al.* \(1998\)](#), for the detection of the 20210G>A mutation. DNA is extracted and amplified using allele-specific primers for the normal and mutant prothrombin alleles. The amplicons produced have different sizes, and are separated by gel electrophoresis. This method allows for complete genotyping of individuals for mutant and normal prothrombin alleles.

SPECIMEN REQUIREMENTS

We require a minimum of 1 milliliter of whole blood. Blood samples must be collected in EDTA (purple topped) tubes.

TRANSPORT

If the specimen is from clinics within UAB or Kirklin Clinic, please call 934-5562 for pickup. If specimens are being sent from some other location, please ship via **UPS**

Overnight Priority. Specimens must be packaged to prevent breakage and absorbent material must be included in the package to absorb liquids in the event that breakage occurs. Also, the package must be shipped in double watertight containers (e.g. a specimen pouch + the shipping companies Diagnostic Envelope). Please contact us (Email – mgl@genetics.uab.edu, Phone – 205-934-5562) prior to sample shipment and provide us with the date of shipment and the tracking number of the package.

TURN AROUND TIME: 7-10 working days

CPT CODES AND PRICES: \$170 USD

83891, 83894 (x2), 83900 (x2), 83912

Please note that prices listed correspond to institutional rates; please contact the lab for insurance rates.

Payment may be made by Money order, Cashier's check, VISA or MasterCard. We will also bill requesting institutions. Please call for more information.

REQUIRED FORMS

[Test Requisition Form](#)

Note: Requests for Molecular Genetic testing for Factor II will **not** be accepted for the following reasons:

- No label (patients full name and date of collection) on the specimens
- No referring physician's or genetic counselor's names and addresses
- No billing information if being paid for by an institution
- No informed consent

For more information, test requisition forms, or sample, please call: 205-934-5562.

REFERENCES

[Héazard, N., Cornillet-Ldfevre, P., Gillot, L., Potron, G., Nguyen, P., \(1998\) Multiplex ASA PCR for a simultaneous determination of Factor V Leiden gene, G>A 20210 Prothrombin gene and C>T 677 MTHFR gene mutations. *Thromb. Haemost.* 79: 1054-1055.](#)

[Reich LM, Bower M, Key NS. \(2003\) Role of the geneticist in testing and counseling for inherited thrombophilia. *Genet Med.* 2003 May-Jun;5\(3\):133-43.](#)

[Varga EA, Moll S., \(2004\) Cardiology patient pages. Prothrombin 20210 mutation \(factor II mutation\). *Circulation.* Jul 20;110\(3\):e15-8](#)