

Prothrombin G20210A

Description:

The Factor II (Prothrombin) G20210A variant is the second-most common cause of inherited thrombosis (after factor V Leiden thrombophilia). This variant causes elevated levels of prothrombin, which mildly increases the risk of venous thrombosis. An increased risk of venous thrombosis at certain unusual sites such as the cerebral, mesenteric, portal, or hepatic veins may also be associated with this variant. The G20210A variant results in approximately 30% higher levels of prothrombin, which can cause a mild hypercoagulable condition associated with deep vein thrombosis. Heterozygosity for the G20210A variant results in a 2 to 5-fold higher risk of thrombosis. The risk of thrombosis associated with homozygosity for the G20210A variant is not well defined, but is thought to be higher than the risk associated with heterozygosity. Combined heterozygosity for Factor V Leiden and Prothrombin G20210A results in up to a 20-fold increased risk of thrombosis. The population frequency of Prothrombin G20210A variant is between 1% and 4% in Caucasians and about 0.2% in African-Americans.

Multiple professional societies recommend testing for Prothrombin only when results would affect clinical management.

Indications:

When results would affect clinical management, consider testing with the following indications:

- Unprovoked first or recurrent venous thromboembolism, especially when it manifests as deep vein thrombosis or pulmonary embolism
- Venous thromboembolism at an unusual site (cerebral, mesenteric, portal, or hepatic veins)
- Family members known to have Prothrombin G20210A or recurrent thrombosis

- Females with venous thromboembolic event during pregnancy or in association with use of oral contraceptives or hormone replacement therapy

Specimen:

At least 3 mLs of whole blood in lavender top (EDTA) tube. Label tube with patient's name, birth date, and date of collection. Phlebotomist must initial tube to verify patient's identity.

Testing Methodology:

Allelic Discrimination TaqMan Assay (Applied Biosystems) is used to determine the genotype at the Prothrombin locus. End-products are analyzed using the ABI 7500 Real-Time PCR System for genotype detection.

*Analysis of the Prothrombin 20210G>A variant is also offered as part of our Thrombophilia Panel which includes the Factor V Leiden variant.

Sensitivity:

This test methodology detects >99% of instances of the variant.

Turn-Around Time:

7 days

CPT Codes:

- 81240

Please call 1-866-450-4198 for current pricing, insurance preauthorization or with any billing questions.

Results:

Each test report includes a detailed interpretation of the genetic findings, the clinical significance of the result, and specific recommendations for the clinical management and additional testing, if warranted. Results will be reported to the referring physician or health care provider as specified on the test requisition form.

Shipping Instructions:

Please enclose test requisition with sample.
All information must be completed before sample can be processed.

Place samples in styrofoam mailer and ship at room temperature by overnight Federal Express to arrive Monday through Saturday.

Ship to:

Cytogenetics and Molecular Genetics Laboratories
3333 Burnet Avenue NRB 1042
Cincinnati, OH 45229
513-636-4474

References:

ACOG. ACOG Practice Bulletin No. 138: Inherited thrombophilias in pregnancy. *Obstet Gynecol.* 2013b; 122:706-17.

Arachchillage, D. J., Mackillop, L., Chandratheva, A., Motawani, J., MacCallum, P., & Laffan, M. (2022). Thrombophilia testing: A British Society for Haematology guideline. *British journal of haematology*, 198(3), 443–458.

Chong LY, Fenu E, Stansby G, Hodgkinson S. Management of venous thromboembolic diseases and the role of thrombophilia testing: summary of NICE guidance. *BMJ* 2012;344:e3979.

Dentali F, Ageno W, Bozzato S, Malato A, Gianni M, Squizzato A, Prisco D. Role of factor V Leiden or G20210A prothrombin mutation in patients with symptomatic pulmonary embolism and deep vein thrombosis: a meta-analysis of the literature. *J Thromb Haemost.* 2012;10:732–7.

EGAPP Working Group. Recommendations from the EGAPP Working Group: routine testing for Factor V Leiden (R506Q) and prothrombin (20210G>A) mutations in adults with a history of idiopathic venous thromboembolism and their adult family members. *Genet Med.* 2011;13:67–76.

Middeldorp, S., Nieuwlaet, R., Baumann Kreuziger, L., Coppens, M., Houghton, D., James, A. H., Lang, E., Moll, S., Myers, T., Bhatt, M., Chai-Adisaksopha, C., Colunga-Lozano, L. E., Karam, S. G., Zhang, Y., Wiercioch, W., Schünemann, H. J., & Iorio, A. (2023). American Society of Hematology 2023 guidelines for management of venous thromboembolism: thrombophilia testing. *Blood advances*, 7(22), 7101–7138.

Rosendaal FR, Reitsma PH. Genetics of venous thrombosis. *J Thromb Haemost.* 2009;7 Suppl 1:301–4
Stevens SM, Woller SC, Bauer KA, Kasthuri R, Cushman M, Streiff M, Lim W, Douketis JD. Guidance for the evaluation and treatment of hereditary and acquired thrombophilia. *J Thromb Thrombolysis.* 2016;41:154–64.