Factor II (Prothrombin G20210A)

The Factor II (Prothrombin) G20210A assay allows the detection and genotyping of a single point mutation (G to A at position 20210) of the human Factor II gene from DNA isolated from human whole peripheral blood. The test is performed on the LightCycler instrument utilizing polymerase chain reaction (PCR) for the amplification of Factor II DNA recovered from clinical specimens and fluorogenic target-specific hybridization for the detection and genotyping of the amplified Factor II DNA.

Detection and genotyping of the Factor II (Prothrombin) G20210A mutation aids in the evaluation of patients with suspected thrombophilia. Inherited thrombophilia predispose an individual to thrombotic events such as venous thrombosis, the third most common cardiovascular disease. Activated protein C (APC) resistance is regarded as the most prevalent coagulation abnormality associated with venous thrombosis. Patients testing positive for APC resistance or the Factor V Leiden mutation should be considered for molecular genetic testing for the other most common thrombophilias with overlapping phenotype, for which testing is available at present [*i.e.*, the Factor II (Prothrombin) G20210A variant]. The Factor II 20210A mutation is present in 1-2% of the general population and its involvement in venous thromboembolism is well established.

How are the results reported?

The results for Factor II (Prothrombin 20210A) testing are reported as one of three possible results:

NORMAL: A **Normal** result means that the Factor II (Prothrombin 20210A) allele (or mutation) was not detected on either version/copy of the Factor II gene.

HETEROZY: This result (Heterozygous) indicates that one copy of the Factor II (Prothrombin) allele has been identified with the 20210A mutation.

HOMOZYGO: This result (Homozygous) indicates that both copies of the Factor II (Prothrombin) allele have been identified with the 20210A mutation.

Important Additional Notes:

- This assay is a germline genetic assay and should only be ordered once on any particular patient. The Prothrombin 20210A genotype is constitutional and does not change.

- The Prothrombin 20210A assay is typically ordered in conjunction with the Factor V Leiden mutational assay and other markers of hypercoagulability, in patients with thrombosis.

References:

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2. Wisotzkey, J.D., P. Bayliss, E. Rutherford and T. Bell. *Placental Genotyping of the Factor V Leiden, Prothrombin 20210A and the Methylenetetrahydrofolate Reductase (MTHFR) C677T Alleles in IUGR Pregnancies (Letter)*. **Thrombosis and Haemostasis,** 81:844-845, 1999
3. Wisotzkey, J.D., Bell, T. and Monk, J.S. *Simultaneous Polymerase Chain Reaction Restriction Fragment Length Polymorphism (PCR-RFLP) Identification of the Factor V Leiden Allele and the Prothrombin 20210A Mutation.* **Diagnostic Molecular Pathology**, 7:180-183, 1998.
4. Spector et al. Technical standards and guidelines: Venous thromboembolism (Factor V Leiden and prothrombin 20210G>A testing): A disease-specific supplement to the standards and guidelines for clinical genetics laboratories. **Genet Med**, 7:444-453, 2005.

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