

UCLA DIAGNOSTIC MOLECULAR PATHOLOGY LABORATORY
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Prothrombin 20210A Variant

↑CPT

83890; 83898; 83896 (x2); 83903; 83912

↑Laboratory

Molecular Pathology

↑Turnaround Time

3-14 days

↑Specimen

Blood

↑Volume

5 mL whole blood

↑Container

Lavender top (EDTA) tube

↑Storage Instructions

All specimens should be sent to the Laboratory immediately after collection, preferably by overnight delivery. Specimens should be kept at room temperature or refrigerated but not frozen.

↑Reference Range

No 20210A variant detected

↑ Use

Deep vein thrombosis is a common disease, with an annual incidence in the general population of approximately 1 per 1000. Several hereditary risk factors have been identified by now. One of them is a genetic variation in the 3'-untranslated region of the prothrombin gene. A transition of G to A at nucleotide position 20210 was found in 18% of selected patients with a personal and family history of venous thrombosis; in 6.2% of unselected consecutive patients with a first, objectively confirmed episode of a deep-vein thrombosis; and in 2.3% of healthy control subjects. Carriers of the 20210A allele have higher plasma prothrombin levels than controls with the normal 20210G genotype and have a 2.8-fold increased risk of venous thrombosis.

↑ Limitations

Whether or not the 20210A variant is detected, it does not rule out the possibility that other thrombophilic mutations and variants could (also) be present, as there are several other heritable causes of thrombophilia, all of which may interact in a synergistic fashion.

↑ Methodology

The Roche Factor II (Prothrombin) G20210A Kit is performed on the LightCycler® Instrument utilizing polymerase chain reaction (PCR) for the amplification of Factor II DNA recovered from clinical samples. A 165 bp fragment of the Factor II gene is amplified from human genomic DNA using specific primers. The amplicon is detected by fluorescence using a specific pair of hybridization probes. The hybridization probes are also used to determine the genotype by performing a melting curve analysis after the amplification cycles are completed.