

Factor II Prothrombin 20210 G>A Mutation Analysis

For in vitro diagnostic use

CERNER ORDERABLE

PT Gene Mutation PCR; PTMTPCR 7360

CPT CODE

81240

CLINICAL UTILITY

A prothrombin mutation is the second most inherited clotting abnormality, present in approximately 2% of the general population. This mutation results in increase production of the protein which can active thrombin and increase clot formation. The Factor II Prothombin 20210 G>A gene mutation affects approximately 18% of those with recurrent familial venous thrombosis, and there is a 3-fold increased risk of venous thrombosis among people heterozygous for the 20210 G>A mutation of the prothrombin gene. Detection and genotyping of the prothrombin mutation aids in prevention and treatment plans for blood clots¹.

METHODOLOGY

PCR/ Melting Curve Analysis

SPECIMENS

Peripheral blood drawn in a 3 mL or 6 mL EDTA or Sodium citrate tube. Minimum acceptable volume is 2 mL.

For pediatric patients, an EDTA micro container may be used. Minimum acceptable pediatric volume is 500 µL. Do not spin.

SPECIMEN STABILITY and SHIPPING

Store and ship whole blood specimen refrigerated. Do not spin.

CAUSES FOR REJECTION

Clotted or contaminated sample; if collected in any other anticoagulant.

SPECIFICITY

Factor II (Prothrombin) G20210A mutation is the only mutation identified in this assay.

ASSAY RESULTS

Genotype differentiation between wildtype (normal), heterozygous, or homozygous mutation.

TURNAROUND TIME

Thursday, 7 days

1. Reference information can be found in the Indiana University Health Molecular Assay Procedures.