

## **Factor II (Prothrombin) Gene Mutation, Molecular Methods**

**Test Code:** FACII(individual order) or HCOG (Hyper-coagulation Panel)

**Use:** Factor II (Prothrombin) test is a diagnostic test for the detection and genotyping of the Factor II (Prothrombin) G20210A mutation as an aid to diagnosis in the evaluation of patients with suspected thrombophilia.

**Clinical Significance:** Genetic alterations predisposing to thrombosis are common in the general population. The Factor II (Prothrombin) gene mutation is one of the genetic risk factors for thrombosis and is associated with elevated prothrombin levels. Prothrombin gene mutation is the second most common genetic defect for inherited thrombosis.

**Methodology:** Extraction of genomic DNA from whole blood is followed by amplification using polymerase chain reaction (PCR) and detection using a melting curve analysis. During the melting curve analysis, fluorescence is continuously monitored while the temperature is increased. The increasing temperature will result in a dissociation of the amplified product at a specific temperature (melting temperature,  $T_M$ ). At the  $T_M$ , the fluorescence properties decrease rapidly. In the presence of a mutation, the resulting  $T_M$  will be lower than the wild type.

**Normal Range:** No Mutation Detected

**Reportable Range:** Results for the Factor II (Prothrombin) test are reported as homozygous mutation (two copies for the mutation), heterozygous mutation (one copy of the mutant gene and one copy of the normal (wild type) gene), or no mutation detected (two copies of the normal (wild type) gene).

**Assay Availability:** Batched once weekly on Tuesday

**Results Reported:** 7 days

**Specimen:** Whole Blood: 1 lavender top (EDTA) tube  
Specimens should be labeled with patient name, medical record number, date and time of collection, and then sent at room temperature to the laboratory. Specimens used for testing in other departments are unacceptable due to possible contamination.

**Volume:** 4 mL (minimum volume: 1 mL)

**Storage:** If not tested immediately, store whole blood refrigerated.

**Causes for Rejection:** Heparinized specimens; Quantity not sufficient for analysis; specimen grossly contaminated; frozen whole blood specimens; specimen leaky or tube broken.

**Laboratory Contact:** For further information, please call the Molecular Diagnostics Laboratory at (501) 526-6439.

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