Prothrombin (Factor II) G20210A Mutation

CPT Code: 81240
Order Code: C607
ABN Requirement: Yes
Synonyms: Prothrombin
Specimen: EDTA Whole Blood
Volume: 1.0 mL
Minimum Volume: 0.5 mL
Container: EDTA (Lavender Top Tube)

Collection:
1. Collect and label sample according to standard protocols.
2. Gently invert tube 8-10 times immediately after draw. DO NOT SHAKE.
3. Do not centrifuge.

Special Instructions: Please provide a separate specimen tube for molecular testing.

Transport: Store EDTA whole blood at 2°C to 8°C after collection and ship the same day per packaging instructions provided with the Cleveland HeartLab, Inc. shipping box.

Stability:
- Ambient (15-25°C): 48 hours
- Refrigerated (2-8°C): 7 days
- Frozen (-20°C): 1 year
- Deep Frozen (-70°C): 1 year

Causes for Rejection: Specimens other than EDTA whole blood; improper labeling; samples not stored properly; samples older than stability limits

Methodology: Polymerase Chain Reaction (PCR); Melt Curve Analysis

Turn Around Time: 7-10 days

Reference Range:
- Negative: The patient is negative of Prothrombin G20210A mutation.

Clinical Indication: The Factor II test is indicated for patients with early onset, recurrent, or unexplained thrombotic events

Clinical Significance: Prothrombin Gene Mutation 20210A has been described to be associated with thrombotic risk. The most common mutation is a G to A transition in position 20210 in the 3’ untranslated region. This mutation is associated with an increase in plasma prothrombin level.
Prevalence of the mutation is ~2% in the general population. It is an autosomal dominant condition and most of affected patients are heterozygous. Homozygotes are rare and they demonstrate early onset of thrombophilia and increased severity. Increased severity can be also seen in patients who are heterozygous for both the Factor V Leiden and prothrombin gene mutations.