Interpretive Data:

Background Information for Prothrombin (F2) c.*97G>A (G20210A) Pathogenic Variant:

Characteristics: The Factor II c.*97G>A (G20210A) pathogenic variant is a common genetic risk factor for venous thrombosis associated with elevated prothrombin levels leading to increased rates of thrombin generation and excessive growth of fibrin clots. The expression of Factor II thrombophilia is impacted by coexisting genetic thrombophilic disorders, acquired thrombophilic disorders (e.g., malignancy, hyperhomocysteinemia, high factor VIII levels), and circumstances including: pregnancy, oral contraceptive use, hormone replacement therapy, selective estrogen receptor modulators, travel, central venous catheters, surgery, and organ transplantation.

Incidence: Approximately 2 percent of Caucasians and 0.3 percent of African Americans are heterozygous; homozygosity occurs in 1 in 10,000 individuals.

Inheritance: Incomplete autosomal dominant.

Penetrance: The risk of thrombosis is increased 2-4 fold for heterozygotes and further increased for homozygotes.

Cause: Homozygosity or heterozygosity for F2 c.*97G>A (G20210A).

Pathogenic Variant Tested: F2 c.*97G>A (G20210A).

Clinical Sensitivity for Venous Thrombosis: Approximately 10 percent.

Methodology: Polymerase chain reaction and fluorescence monitoring.

Analytical Sensitivity and Specificity: 99 percent.

Limitations: Diagnostic errors can occur due to rare sequence variations. F2 gene variants, other than c.*97G>A (G20210A), will not be detected.

See Compliance Statement C: www.aruplab.com/CS

HOTLINE NOTE: Remove information found in the Reference Interval field.