

Factor V Leiden Mutation Analysis

For in vitro diagnostic use

CERNER ORDERABLE

F-V DNA LEIDEN BLD PCR; LEIDEN 3397

CPT CODE

81241

CLINICAL UTILITY

Detection and genotyping of the Factor V Leiden (G1691A) mutation by PCR is used as an aid to diagnosis in the evaluation of patients with suspected thrombophilia. Inherited thrombophilia predisposes an individual to thrombotic events such as venous thrombosis, the third most common cardiovascular disease. Activated protein C (APC) resistance is regarded as the most prevalent coagulation abnormality associated with venous thrombosis. A point mutation at of the Factor V gene renders the Factor V protein partially resistant to inactivation by APC. Genetic analysis has demonstrated that this mutation, which has a relatively high prevalence in the general population (~ 5% in Caucasians), accounts for 85% to 95% of APC resistance cases. This abnormality is associated with an increased risk of venous thromboembolism (5-10 fold in heterozygotes and 50-100 fold in homozygotes), juvenile stroke, myocardial infarction in juvenile women, and traumatic conditions¹.

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

Does not allow the differentiation of the rare silent mutations A1692C, G1689A and A1696G.

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.