BACKGROUND:
Poor plasma anticoagulant response to activated protein C (APC resistance) is the most commonly observed inherited defect associated with thrombotic risk.\textsuperscript{1} The transition of guanine (G) to adenine (A) at position 1691 of the factor V gene causes substitution of glutamine for arginine at amino acid 506, resulting in Factor V Leiden.\textsuperscript{2} This substitution renders Factor V Leiden resistant to inactivation by activated protein C, and is the defect underlying the majority of cases of APC resistance. Heterozygous carriers of the Factor V G1691A allele have approximately an 8 fold increased risk of venous thrombosis, while individuals homozygous for Factor V G1691A have an estimated increased risk of 91 fold compared to normal individuals.\textsuperscript{3}

REASONS FOR REFERRAL:
• Evaluation of individuals with thrombosis or a family history of thrombosis.
• Evaluation of patients with complications of pregnancy, such as pre-eclampsia or late fetal loss.\textsuperscript{4}

METHOD:
Gene amplification followed by detection with sequence-specific FRET hybridization probes. As this is a genetic test, genetic counseling may be appropriate. Informed consent is recommended, and is required in New York. Consent forms are available upon request.

LIMITATIONS:
Samples may be taken from patients on any form of anticoagulation. Since the test is performed on DNA derived from white blood cells, severe leukopenia may compromise DNA extraction, and in marrow transplant patients, DNA extracted will reflect the donor genetic traits rather than those of the patient. Specificity and sensitivity for detection of G1691A is > 99%. Specificity may be affected by mutations in the PCR priming sites.

NORMAL VALUES:
Normal sequence at factor V 1691. Abnormal results will be interpreted as heterozygous or homozygous.

SPECIMEN REQUIREMENTS:
5 ml EDTA whole blood, collected and shipped at room temperature. Sample must be less than 1 month old when received by our laboratory.
SHIPPING REQUIREMENTS:
Place the specimen and the test requisition form into plastic bags and seal. Insert into a Styrofoam container; place into a sturdy cardboard box, tape securely and ship by an overnight carrier. Ship the package in compliance with your overnight carrier guidelines. Label with the following address:

Client Services/Hemostasis Reference Laboratory
BloodCenter of Wisconsin
638 N. 18 St.
Milwaukee, WI 53233
Phone: 800-245-3117, ext. 6250

TURNAROUND TIME: 3-6 days

CPT CODES: 81241

RELATED PROFILES:

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REFERENCES: