A SCREEN FOR FACTOR V LEIDEN

Factor V Leiden (FVL) is the most common cause of hereditary thrombophilia. The major clinical manifestation is DVT (deep vein thrombosis) with or without pulmonary embolism. The risk is comparably increased for cerebral vein thrombosis. FVL is present in approximately 10-20% of patients with a first VTE event without a family history of VTE. There is also evidence that the FVL mutation may play a role in some cases of unexplained recurrent pregnancy loss. Heterozygous carriers of FVL have been shown to have an overall 3-7 fold increased risk of venous thrombosis, while homozygotes have a 50 to 100-fold risk.

The Activated Protein C Resistance (APCR) Test

The APCR clotting test is an important component of laboratory panels for venous thrombosis. Resistance to activated protein C (APCR) is a relatively frequent finding in patients with unexplained or familial venous thromboembolism (VTE), and is associated with FVL.

APCR is:

- the most common cause of inherited thrombophilia.
- present in 5% of the Caucasian population and accounts for 50% of the inherited thrombophilic cases in this group.
- present in 30-50% of those with recurrent venous thrombosis.

Characteristics of the assay:

- There is excellent and wide discrimination between normal individuals and carriers of the Factor V Leiden mutation.
- The assay we use is not affected by anticoagulation with heparin (standard or low molecular weight) or warfarin.
- There is no interference by Factor VIII and/or lupus anticoagulants as occurred with past coagulation tests of this type.

A normal result rules out the presence of Factor V Leiden, while an abnormal result will be followed up with a PCR assay to confirm the factor’s presence.
Follow-up to an order for Factor V Leiden:

The APCR assay (100% sensitivity; 98% specific) will be performed first as a screen since the majority of these tests are negative, and the screen is far less expensive than a PCR assay.

If the ratio is $\geq 2.5$ no further testing is done as the patient is negative for Factor V Leiden.

If the ratio is $\leq 2.0$ the laboratory will automatically follow up with a confirmatory PCR assay for Factor V Leiden. The PCR test is not only performed for confirmatory measures but also to distinguish between heterozygous and homozygous individuals.

Ratios between 2.1 and 2.4 are indeterminate and should be investigated further. (e.g. Protein C).


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