An individual with a documented thrombotic episode should undergo a complete medical evaluation to rule out conditions associated with thrombophilia not diagnosed by first line testing, eg, nephrotic syndrome, diabetes mellitus, etc. High risk individuals are those with a strong family history of thrombosis and/or those with acquired risk factors, eg, obesity, prolonged immobilization, etc. All non-genetic testing should be repeated in 6 to 12 weeks to reduce the likelihood of false-positives. Some assays are affected by anticoagulants or the acute thrombotic process. Basic blood tests for occult cancer evaluation may include CBC, erythrocyte sedimentation rate, liver and renal function tests, urinalysis, protein electrophoresis, and chest radiography. In patients being evaluated for occult cancer, ultrasound and/or computed tomography (CT) of the abdomen and pelvis may be performed along with the tumor marker tests. Second line testing is for the identification of rare causes of thrombophilia and recommended for individuals with a documented thrombotic episode(s), a strong family history of thrombosis, and negative first line tests. APCR/FVL, indicates activated protein C resistance/factor V Leiden; PTT-LA, partial thromboplastin time-lupus anticoagulant; dRVVT, dilute Russell’s viper venom time; and (i)2-GPI, beta2-glycoprotein I.

This figure was developed by Quest Diagnostics based on references 14-17. It is provided for informational purposes only and is not intended as medical advice. A physician’s test selection and interpretation, diagnosis, and patient management decisions should be based on his/her education, clinical expertise, and assessment of the patient.

Figure. Testing Algorithm for the Diagnosis of Thrombophilia in Individuals with a History of Thrombosis or Those at High Risk