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Short Commentary

To Investigate DVT for Thrombophilic Factors, or Not To

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Thrombophilia is an inherited predisposition to Venous Thromboembolism (VTE) of Deep Vein Thrombosis (DVT), with or without associated Pulmonary Embolus (PE). Ever since the deficiency antithrombin was reported as inherited risk factor for venous thromboembolism, the gamut of test has increased seeking causal association for familial venous thrombosis including F5G1691A (FVR506Q, factor V Leiden) and the F2G20210A gene mutations. In the 1980s and 1990s thrombophilia testing became common in unselected patients and their relatives. According to recent guidelines, testing is clearly not mandatory in situations where the clinical utility of testing is not proven, (clinical utility defined as the ability of a test to influence or alter clinical outcome). However, many clinicians still have used thrombophilia test results to determine clinical management. [1]

Clinicians in developing countries continue showing unprecedented enthusiasm in ordering tests for cases of arterial or venous thrombosis as well as their family members in absence of family history of thrombosis, sometimes without giving any consideration to very important issues such as time since thrombotic event, patient on anticoagulant therapy as well as when an obvious plausible provoking factor already identified.

Venous thromboembolism is commonly encountered by gynecologists, obstetricians, Orthopedicians, surgeons and they do not have the best of knowledge about thrombophilic factors, their limitations, and relevance and this could be a reason for excessive demand for thrombophilic tests. Also the health system in many developing countries is such that the laboratory experts have no active role in advising the clinicians or modifying the test requests from clinicians. The high cost of these tests at questionable benefit is also overlooked.

Thrombophilia testing in patients with a first VTE do not reduce the incidence of recurrence in clinical practice [2]. Thus knowledge of the thrombophilia status of affected patients fails to substantially help clinicians to prevent any recurrence. Most evidences currently suggest that the intensity and duration of anticoagulation treatment after a thrombotic event should not be altered based on most thrombophilic risk factors status [3].

Testing at the time of acute venous thrombosis is not indicated as treatment of acute venous thrombosis is not influenced by test results, immediate testing is not indicated [1]. Test requisitions on individuals after an acute event or initiation of anticoagulant therapy may result in wrong diagnosis of Antithrombin, Protein C, and/ or Protein S deficiencies and Activated Protein C resistance. One third of requests for thrombophilia testing are actually during ongoing anticoagulant therapy [4].

The consequence of rampant family study in VTE, is labeling a carrier state of a genetic thrombophilic defect in perfectly healthy family members. Several negative effects of both psychological and social origin are reported [5,6]. Furthermore, difficulties in obtaining life or disability insurance are frequently encountered by individuals who are known carriers of thrombophilia, regardless of whether they are symptomatic or asymptomatic [5]. Testing of any patient or their relatives generates needless anxiety or indeed promotes false reassurance in those who are reported as being negative for the various tests performed.

To conclude, a more informed decision to test for thrombophilic factors is very important and indiscriminate thrombophilia testing should be discouraged by clinicians.

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