When to perform thrombophilia screening

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Identifying the stroke mechanism is a crucial aspect of secondary prevention. However, no stroke etiology is identified in 30-40% of patients. Cryptogenic stroke is the term used to refer to strokes for which no definite cause can be identified.

Thrombophilia is defined as a predisposition to form clots inappropriately, and can be inherited or acquired. The inherited thrombophilias include deficiency of natural anticoagulants, such as protein C, protein S or antithrombin, and factor V Leiden or prothrombin G20210A gene mutation. The most important acquired thrombophilias are antiphospholipid syndrome and cancer.

Thrombophilia is an important risk factor for venous thromboembolism, but its role in arterial thrombosis is not well defined. The presence of thrombophilia has been accurately investigated in this setting, obtaining controversial results.

Multiple case-control studies and a meta-analysis failed to show an association between inherited thrombophilia and stroke. The only strong association found was with the presence of antiphospholipid antibodies, both in retrospective and prospective studies. Plasma levels of antiphospholipid antibodies were elevated in young adults who suffered a stroke compared with controls, and these patients presented a higher risk of recurrent thrombotic events.

Before performing these tests, all other causes must be ruled out. There are many doubts if the information obtained in thrombophilia screening is sufficiently relevant to change clinical decision, regarding secondary prevention. As a consequence, there is no consensus about the clinical utility and cost effectiveness of thrombophilia screening in arterial thrombosis.