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Opinion Article

Prevalence and Risk Factors of Thrombophilic Disorders in Young Stroke Patients

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DESCRIPTION

Stroke, a leading cause of mortality and disability worldwide, is commonly associated with advancing age and established risk factors such as hypertension, diabetes, and atherosclerosis. However, a significant proportion of stroke cases occur in young individuals, presenting a unique challenge for clinicians. In recent years, researchers have explored the role of thrombophilia disorders in young stroke, shedding light on the underdiagnosed and potentially treatable causes of this devastating condition.

Thrombophilic disorders encompass a group of inherited or acquired conditions that promote abnormal blood clotting, leading to an increased risk of thromboembolic events. These disorders can affect various components of the coagulation cascade, including platelets, endothelial cells, and clotting factors. In young stroke patients, an underlying thrombophilia disorder can contribute to the development of ischemic stroke by promoting the formation of blood clots in cerebral arteries.

Prevalence and risk factors

Thrombophilic disorders are relatively rare in the general population; however, their prevalence is higher among young stroke patients. Studies have indicated that approximately 10%-15% of strokes occurring in individuals under the age of 50 are attributable to thrombophilia. In contrast to traditional stroke risk factors, thrombophilic disorders can be present even in the absence of conventional cardiovascular risk factors.

Role of thrombophilia in young stroke

Thrombophilic disorders contribute to young stroke through multiple mechanisms. The most common mechanism involves the formation of arterial clots due to hypercoagulability. Inherited thrombophilias, such as factor V Leiden mutation, prothrombin gene mutation, and deficiencies of anti-thrombin, protein C, or protein S, have been implicated in promoting arterial thrombosis. Additionally, acquired thrombophilias, including antiphospholipid syndrome and hyperhomocysteinemia, can also increase the risk of stroke in young individuals.

Diagnostic approaches

When investigating thrombophilic disorders in young stroke patients, a comprehensive diagnostic workup is crucial. The evaluation typically involves assessing the patient's clinical history, family history, and laboratory investigations. Essential laboratory tests include screening for common inherited thrombophilias using genetic tests, measuring levels of coagulation factors and natural anticoagulants, and testing for the presence of antiphospholipid antibodies. Furthermore, imaging techniques, such as Magnetic Resonance Imaging (MRI) and Computed Tomography (CT), help identify stroke etiology and rule out other potential causes.

Implications for treatment and prevention

Detecting thrombophilic disorders in young stroke patients has significant implications for treatment and prevention strategies. Anticoagulation therapy, with agents such as warfarin or direct oral anticoagulants, is often recommended for secondary prevention in patients with confirmed thrombophilia-related stroke. This treatment approach helps prevent recurrent thromboembolic events and reduce the risk of long-term disability. However, the duration of anticoagulation therapy should be carefully determined, considering the individual's risk-benefit profile.

In addition to treatment, early identification of thrombophilic disorders in young stroke patients has implications for primary prevention in at-risk individuals. Lifestyle modifications, such as smoking cessation, regular exercise, and healthy dietary habits, should be emphasized to mitigate the impact of traditional risk factors. Moreover, genetic counselling and family screening are crucial to identify asymptomatic individuals at risk and provide appropriate interventions to prevent stroke occurrence.

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Challenges and future directions

Despite the growing recognition of thrombophilic disorders in young stroke, several challenges remain. The diagnostic approach is complex and requires expertise in both neurology and hematology. Additionally, the evidence supporting the optimal management strategies for thrombophilia-related stroke is limited, and further research is needed to define the most effective treatment and prevention modalities. Large-scale prospective studies and collaborative efforts are necessary to enhance our understanding of the pathophysiology, natural history, and outcomes associated with thrombophilic disorders in young stroke patients.