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ORAL PRESENTATION

MTHFR C677T mutation: a controversial aetiology of cerebral venous thrombosis

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Abstract

Introduction: Cerebral venous thrombosis (CVT) is a rare neurological illness that represents 0.5-1% of all strokes and is more common in young women. The major risk factors are prothrombotic conditions, oral contraceptives, pregnancy, malignancy and infections. Headache is the most frequent symptom but seizures, nausea, vomiting and focal deficits can also be present. Imaging is fundamental for its diagnosis.

Case Report: A 57-year-old man with hypertension presented at the emergency room, for the second time, with headache initiated 4 days before with increasing intensity. Initially, headache was frontal with orbital irradiation and then occipital to cervical accompanied by nausea and vomiting. There was no history of trauma or infection. Vital signs, physical and neurological examination were normal. The investigation revealed: blood count, electrolytes, kidney and hepatic function with no abnormalities; D-dimers 0.74 ug/ml and brain computed tomography and magnetic resonance showed CVT of the superior sagittal sinus, straight sinus and right sigmoid sinus and anticoagulation was started. An extensive diagnostic workup was made during hospitalization with no underlying aetiology or risk factor identified. Prothrombotic study was performed 6 months after treatment and showed normal levels of homocysteine, antithrombin III, protein C and S, factor V Leiden, fibrinogen, D-dimers, anti-cardiolipin antibodies and lupus anticoagulant. Genetic testing for mutations in the prothrombin gene (G20210A) and methylenetetrahydrofolate reductase (MTHFR) gene (C677T) revealed homozygous mutation in the last one.

Conclusion: The diagnosis of CVT is still a challenge to physicians due to the great variety and lack of specificity on clinical presentation. Based on the aetiological study, it was assumed that the CVT was due to homozygous mutation of the MTHFR gene although there were normal levels of homocysteine, folate and cobalamin. The authors highlight the diagnostic challenge and emphasize the importance of the workup in a disorder with myriad causes. Although some controversy on literature exists, there are reports where the MTHFR C677T polymorphism was associated with CVT.

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