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Young Stroke Unveiling an MTHFR Mutation: A Case of Hyperhomocysteinemia as a Hidden Culprit

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Abstract

Stroke in young adults is uncommon and often linked to nontraditional risk factors. Hyperhomocysteinemia, frequently due to methylenetetrahydrofolate reductase (MTHFR) mutations, predisposes to endothelial dysfunction and thrombosis. We report a young patient presenting with ischemic stroke without conventional vascular risk factors. Evaluation revealed markedly elevated homocysteine and an MTHFR mutation. Other etiologies were excluded. The patient received standard stroke management plus folate, vitamin B12, and vitamin B6 supplementation, with neurological improvement. This case highlights the importance of considering MTHFR-related hyperhomocysteinemia in young stroke for timely intervention and secondary prevention.

Keywords: NIL

Introduction

Stroke in young adults accounts for a small but clinically significant proportion of ischemic strokes. In the absence of conventional vascular risk factors, uncommon etiologies must be considered. Hyperhomocysteinemia is a recognized prothrombotic and atherogenic state associated with increased risk of arterial and venous thrombosis. Mutations in the methylenetetrahydrofolate reductase (MTHFR) gene, such as C677T or A1298C, impair folate metabolism and elevate homocysteine levels. This case illustrates how hyperhomocysteinemia due to MTHFR mutation can present as ischemic stroke in a young patient.

Case Report

A young 32 year old male presented with acute onset neurological deficit characterized by right sided weakness. There were no conventional stroke risk factors, such as hypertension, diabetes, dyslipidemia, or smoking history. Neuroimaging confirmed acute ischemic stroke. Laboratory investigations revealed markedly elevated plasma homocysteine levels. Genetic analysis identified an homozygous C677T MTHFR mutation . Cardioembolic, vasculitic, and infectious causes were excluded. The patient was treated with standard stroke protocol alongside homocysteine-lowering therapy with folic acid, vitamin B12, and vitamin B6. Secondary prevention with antiplatelet therapy and lifestyle modification was initiated. Neurological recovery was partial on follow-up.

Discussion

Hyperhomocysteinemia is increasingly recognized as an independent risk factor for ischemic stroke, particularly in young patients without traditional vascular risks. Homocysteine contributes to vascular injury through endothelial dysfunction, oxidative stress, and promotion of thrombosis. MTHFR mutations are among the most common genetic causes of elevated homocysteine. Our case emphasizes the clinical importance of screening for homocysteine levels and genetic variants in young stroke patients. Timely diagnosis allows targeted therapy with folate, vitamin B6, and vitamin B12, which can normalize

homocysteine levels and potentially reduce recurrence. Genetic counseling may further aid family risk assessment.

References

1. Homocysteine Studies Collaboration. Homocysteine and risk of ischemic heart disease and stroke: a meta-analysis. JAMA. 2002;288(16):2015-2022.
2. Kim RJ, Becker RC. Association between MTHFR C677T polymorphism and cardiovascular disease: J Thromb Thrombolysis. 2003;16(3):127-136.
3. Kumar J, Das SK, Sharma P. MTHFR gene variants and hyperhomocysteinemia in stroke: a clinical perspective. Neurol India. 2019;67(2):331-338.
4. Saposnik G, Ray JG, Sheridan P, McQueen M, Lonn E. Homocysteine-lowering therapy and stroke risk, severity, and disability: findings from the HOPE 2 trial. Stroke. 2009;40(4):1365-1372.
5. den Heijer M, Lewington S, Clarke R. Homocysteine, MTHFR and risk of venous thrombosis: a meta-analysis. J Thromb Haemost. 2005;3(2):292-299.

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