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YOUNG STROKE UNVEILING THE ENIGMA: HYPERHOMOCYSTEINEMIA AS THE CULPRIT - A COMPELLING CASE REPORT

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ABSTRACT Stroke is a significant cause of morbidity and mortality worldwide. While the majority of strokes occur in older adults with multiple risk factors, stroke can also affect young individuals. Hyperhomocysteinemia is a condition characterized by elevated levels of homocysteine in the blood and is recognized as an independent risk factor for stroke. Presenting a case of a young adult who experienced an ischemic stroke due to severe hyperhomocysteinemia. This case emphasizes the importance of recognizing and treating this potentially reversible risk factor for stroke, particularly in young patients. Early identification and intervention can significantly reduce the risk of recurrent strokes in affected individuals.

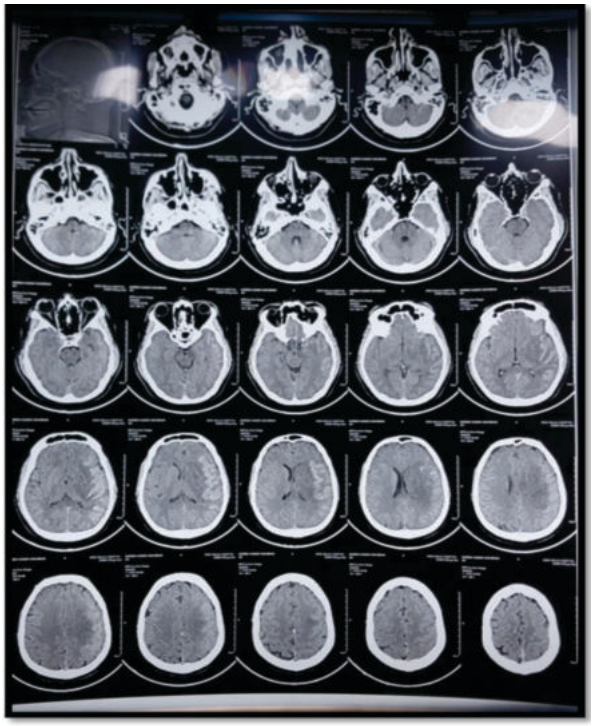
KEYWORDS :

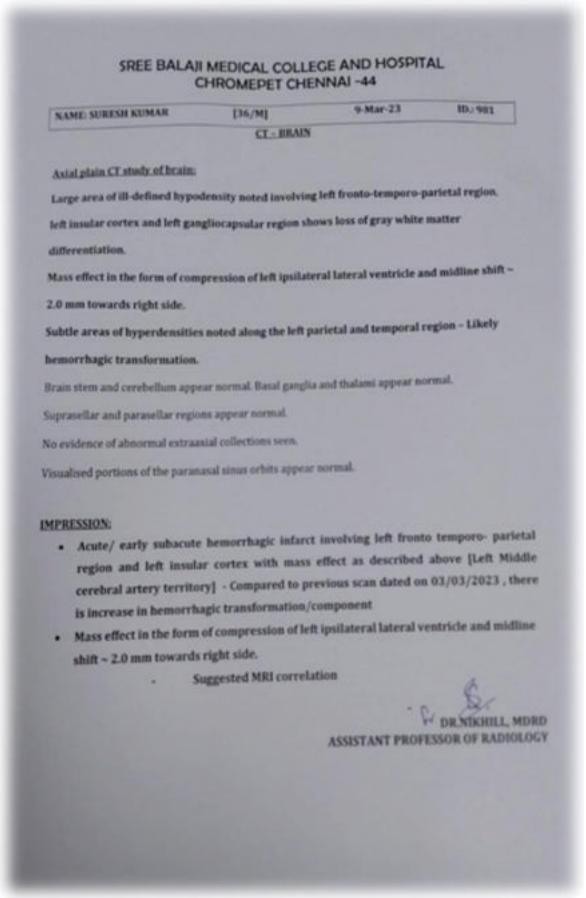
Case Report:

A 32-year-old previously healthy male presented to the emergency department with sudden onset of weakness in his right arm and leg, slurred speech, and facial droop. The symptoms began approximately 3 hours before admission. The patient had no significant medical history, was a non-smoker, and did not use illicit drugs. Family history was unremarkable for stroke or other neurological disorders.

On examination, the patient was conscious but appeared anxious. Neurological examination revealed right-sided hemiparesis, dysarthria, and facial weakness. The National Institutes of Health Stroke Scale (NIHSS) score was 8, indicating a moderate to severe stroke.

Computed tomography (CT) of the brain ruled out any hemorrhage and confirmed an ischemic stroke. The patient was immediately started on thrombolytic therapy with tissue plasminogen activator (tPA) within the appropriate time window.





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CT - BRAIN

Axial plain CT study of brain:

Large area of ill-defined hypodensity noted involving left fronto-temporo-parietal region, left insular cortex and left ganglionocapsular region shows loss of gray white matter differentiation.

Mass effect in the form of compression of left ipsilateral lateral ventricle and midline shift ~ 2.0 mm towards right side.

Subtle areas of hyperdensities noted along the left parietal and temporal region ~ Likely hemorrhagic transformation.

Brain stem and cerebellum appear normal. Basal ganglia and thalami appear normal.

Supraellar and parasellar regions appear normal.

No evidence of abnormal extraaxial collections seen.

Visualised portions of the paranasal sinus orbits appear normal.

IMPRESSION:

- Acute/ early subacute hemorrhagic infarct involving left fronto temporo- parietal region and left insular cortex with mass effect as described above [Left Middle cerebral artery territory] - Compared to previous scan dated on 03/03/2023 , there is increase in hemorrhagic transformation/component
- Mass effect in the form of compression of left ipsilateral lateral ventricle and midline shift ~ 2.0 mm towards right side.

Suggested MRI correlation

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Further investigations included blood tests to evaluate potential risk factors for stroke in a young patient. The lipid profile was within normal limits, and there were no signs of hypercoagulability. However, the serum homocysteine level was markedly elevated at 40 □mol/L (normal range: 5-15 μmol/L).

To identify the underlying cause of the hyper homo cysteinemia, additional tests were performed. Folate, vitamin B12, and vitamin B6 levels were found to be normal. The patient's methylenetetrahydrofolate reductase (MTHFR) gene was sequenced, and a homozygous mutation (C677T) was detected, further confirming the diagnosis of severe

hyperhomocysteinemia due to MTHFR gene mutation.

Treatment and Outcome:

The patient was started on high-dose folic acid and vitamin B12 supplementation to lower the homocysteine levels and reduce the risk of recurrent stroke. Additionally, lifestyle modifications, including a diet rich in folate and B vitamins, were recommended.

Physical therapy and rehabilitation were initiated to aid in the recovery of motor function and speech. The patient made gradual progress during the hospital stay, and his NIHSS score decreased to 2 at discharge.

Follow-up assessments at 3 and 6 months showed significant improvement in neurological deficits, and the patient remained compliant with the prescribed medications and lifestyle modifications.

DISCUSION:

This case highlights the importance of considering hyperhomocysteinemia as a potential cause of stroke, even in young individuals without traditional risk factors. The homozygous MTHFR gene mutation led to severe hyperhomocysteinemia in our patient, resulting in an ischemic stroke.

Genetic predisposition to hyperhomocysteinemia can be identified through genetic testing, enabling targeted interventions to reduce the risk of stroke and other cardiovascular events. Early diagnosis and appropriate management with vitamin supplementation can significantly improve outcomes in affected individuals.

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