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Original Research Paper

Internal Medicine

"MOYAMOYA DISEASE AS A RARE CAUSE OF ISCHEMIC STROKE IN A YOUNG ADULT: A CASE REPORT"

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ABSTRACT **Background:** Moyamoya disease is a rare, progressive cerebrovascular disorder characterized by stenosis or occlusion of the intracranial internal carotid arteries and their proximal branches, leading to the development of abnormal collateral vessels. It is an uncommon but important cause of stroke in young individuals. **Case Presentation:** We report a case of a young adult male presenting with acute onset left-sided weakness, recurrent transient ischemic attacks, and speech disturbance. Neuroimaging revealed bilateral internal carotid artery stenosis with characteristic collateral formation consistent with Moyamoya disease. **Conclusion:** Moyamoya disease should be considered in young patients presenting with ischemic stroke without conventional vascular risk factors. Early diagnosis using advanced imaging modalities is crucial for appropriate management and prevention of recurrent cerebrovascular events.

KEYWORDS : Moyamoya disease, Young stroke, Cerebral infarction, Internal carotid artery stenosis

INTRODUCTION

Moyamoya disease is a rare, chronic, progressive cerebrovascular disorder characterized by bilateral stenosis or occlusion of the terminal portions of the internal carotid arteries and their proximal branches. The progressive reduction in cerebral blood flow leads to the development of fragile collateral vessels at the base of the brain, which on angiographic imaging produce the characteristic "puff of smoke" appearance from which the disease derives its name. Although initially described in East Asian populations, Moyamoya disease is increasingly recognized worldwide across diverse ethnic groups.Clinically, Moyamoya disease exhibits a bimodal age distribution, with one peak in childhood and another in early adulthood. Children commonly present with transient ischemic attacks or ischemic strokes, whereas adults may present with either ischemic or hemorrhagic cerebrovascular events. Recurrent ischemic episodes, headaches, seizures, and cognitive impairment may also occur due to chronic cerebral hypoperfusion.Early diagnosis of Moyamoya disease is critical, as the condition is progressive and associated with a high risk of recurrent cerebrovascular events if left untreated. Magnetic resonance imaging combined with magnetic resonance angiography serves as a non-invasive and reliable diagnostic modality, especially in resource-limited settings. Recognition of this entity is essential in young patients presenting with stroke, as timely medical management and consideration of surgical revascularization can significantly improve neurological outcomes.

We report this case to highlight Moyamoya disease as an important and often underdiagnosed cause of ischemic stroke in young adults, emphasizing the need for heightened clinical suspicion and appropriate neuroimaging in patients presenting with recurrent or unexplained cerebrovascular events.

Case Report

Clinical Profile

An 18-year-old male presented with sudden onset weakness of the left upper and lower limbs associated with difficulty in speech. He had a history of recurrent episodes of transient limb weakness and speech disturbances over the past year, each episode lasting for a few minutes and resolving spontaneously. There was no history of hypertension, diabetes mellitus, cardiac illness, smoking, or substance abuse.

Examination

On examination, the patient was conscious and fully oriented.

Pulse: 88/min

Blood Pressure: 150/95 mmHg

SpO2: 98% on room air

Neurological examination revealed reduced power (3/5) in the left upper and lower limbs. Reflexes were exaggerated on the left side with an extensor plantar response. No cranial nerve involvement, meningeal signs, or sensory deficits were noted. Fundoscopic examination was normal.

Investigations

MRI Brain: Revealed bilateral parietal cortical and subcortical infarcts.

MR Angiography: Showed severe stenosis of bilateral internal carotid arteries with extensive collateral vessel formation, suggestive of Moyamoya disease.

Routine Blood Investigations: Within normal limits.

ECG and Echocardiography: Normal.

Differential Diagnosis

Primary cerebral vasculitis
Secondary vasculitis (systemic lupus erythematosus, antiphospholipid antibody syndrome)
Carotid or vertebral artery dissection
Sickle cell disease–related vasculopathy
Cardioembolic stroke (patent foramen ovale, valvular heart disease)
Fibromuscular dysplasia
Mitochondrial encephalopathy (MELAS)
Reversible cerebral vasoconstriction syndrome

Treatment And Outcome

The patient was managed conservatively with antiplatelet therapy and antihypertensive medications. Neurosurgical consultation was obtained, and the patient was advised for long-term follow-up with consideration for revascularization procedures if symptoms recur.

DISCUSSION

Moyamoya disease is an important yet underdiagnosed cause of ischemic stroke in young patients. The progressive nature of arterial stenosis leads to recurrent ischemic events. MR angiography plays a pivotal role in diagnosis, especially in resource-limited settings. Medical management aims at preventing ischemic events, while surgical revascularization remains the definitive treatment in selected cases.

Diagnostic Challenges

Diagnosing Moyamoya disease in young patients poses significant challenges due to its rarity and nonspecific clinical presentation. Early symptoms often manifest as transient ischemic attacks or subtle neurological deficits, which may be

misattributed to more common etiologies of young-onset stroke such as cerebral vasculitis, arterial dissection, or cardioembolic events. Additionally, routine neuroimaging may demonstrate infarcts without clearly identifying the underlying vasculopathy. Definitive diagnosis requires a high index of suspicion and dedicated vascular imaging, such as magnetic resonance angiography, to demonstrate bilateral internal carotid artery stenosis with characteristic collateral formation. Delayed recognition may lead to recurrent ischemic events and increased neurological morbidity.

CONCLUSION

Young patients presenting with ischemic stroke and recurrent transient ischemic attacks should be evaluated for rare etiologies such as Moyamoya disease. Early diagnosis and appropriate management are essential to prevent recurrent strokes and improve long-term outcomes.

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