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Internal Medicine

A CASE REPORT OF MOYAMOYA DISEASE – THE CULPRIT OF LEFT SIDED HEMIPARESIS IN A YOUNG FEMALE

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ABSTRACT Moyamoya disease is a rare progressive cerebrovascular disease characterized by stenosis of bilateral internal carotid artery with the formation of characteristic, abnormal, fragile collaterals seen as "puff of smoke" appearance on angiography. It occurs predominantly in Japanese individuals but is now reported across all races with bimodal age distribution and varying clinical manifestations. Therefore, Moyamoya disease has been underrecognized as a cause of ischemic and haemorrhagic strokes. At this time, there is no known cure, and existing treatment options are controversial with surgical intervention being more beneficial than medical management. We present a case of 27-year-old diabetic and hypertensive female patient who presented with left sided hemiparesis. She was diagnosed with Right MCA territory acute non haemorrhagic infarct with bilateral ICA,ACA and right MCA stenosis. As there was a history of consumption of oral contraceptive pills initial etiology was considered to be the hypercoagulable state. However the atypical presentation with B/L ICA and ACA stenosis with right MCA stenosis led to Digital Subtraction Angiography (DSA) being done that was diagnostic of Moyamoya disease. Surgical intervention was advised however the patient opted for conservative medical management with antiplatelet and statin. Patient showed clinical improvement in terms of consciousness and was vitally stable on discharge. This case highlights the importance of considering Moyamoya disease in the differential diagnosis of young patients presenting with acute onset neurological symptoms.

KEYWORDS :

INTRODUCTION:

First reported in 1957 in Japan, Moyamoya disease is a rare idiopathic cerebrovascular disease characterized by progressive stenosis of Internal Carotid Arteries (ICA) with subsequent formation of abnormal fragile collaterals at the base of the brain known as Moyamoya vessels. These collaterals are responsible for the characteristic 'puff of smoke' appearance on the digital subtraction angiography (DSA) [1,2]. Moyamoya syndrome includes patients with Moyamoya angiographic findings who also have an associated condition – hematological, vasculitis, genetic or others.

It is more common in East Asian countries with highest prevalence in Japan, China and Korea with annual incidence of Moyamoya being 0.35 to 0.94 per 1,00,000 population. There is a slight female to male preponderance with ratio of 1.9 [3]. The disease has a bimodal distribution with peak at 10 years and then 30-40 years of age [3].

Moyamoya disease has a varying clinical presentation with either TIA and Ischemic stroke or cerebral hemorrhage. Treatment is mainly surgical revascularization. It is a rare cause of stroke in young individuals and is often missed due to limited knowledge about the disease. Here we present a case of 27 year old female patient presenting with left sided hemiparesis who was diagnosed with Moyamoya disease on DSA.

Case Report:

A 27 year old female patient presented to the ER with the chief complain of left sided upper limb and lower limb weakness associated with inability to speak, decreased consciousness and inability to look to the right side since 1 day. There was no history of fever, headache, nausea or vomiting, loss of consciousness, trauma or seizure episode.

The patient had a history of menorrhagia since 1 month for which she had been prescribed Oral Contraceptive Pills and Tranexamic Acid since 15 days. She is a known case of Type 1 Diabetes on insulin since 12 years. She had recently been diagnosed with Hypertension since 15 days and started on Anti-hypertensive medication for the same.

No history of alcohol consumption or smoking; and no significant family history.

On arrival to the hospital, the patient was drowsy but easily arousable; her vitals were - Temperature: 37°C , Blood pressure: 152/100 mm Hg, Heart rate: 90/min, Respiratory rate: 20/min, and oxygen saturation was 97% on room air, with random blood glucose: 520 mg/dl.

On CNS examination – Patient was drowsy but easily arousable; Tone was decreased in Left U/L and L/L; Power was 0/5 in Left U/L and L/L; Left knee and ankle deep tendon reflex were absent with an extensor plantar on the left side. On right side no significant findings were present.

MRI Brain Plain with MRI Head and Neck Angiography was done which was suggestive of Large acute non haemorrhagic infarct involving right frontal lobe, insular cortex and peri insular parieto-temporal lobes and small discrete acute infarct in left parafalcine frontal lobe. MRI Head and Neck Angiography showed complete occlusion of bilateral ICA and ACA with occlusion of right MCA as well [Figure 1 and 2].

MR venogram was unremarkable.

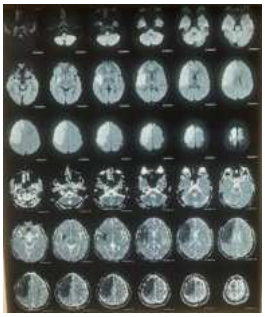

Figure 1 – MRI showing Right MCA territory infarct.

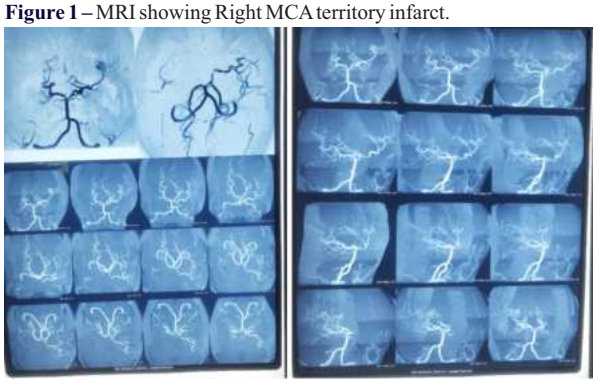

Figure 2 – MRI Head And Neck Angiography showing bilateral ICA stenosis.

Patient was admitted in ICU. Single antiplatelet and statin with prophylactic dose of anticoagulation and antiepileptic were started as per the Neurophysician advise.

In order to identify the cause of stroke in this young patient entire thrombophilia work up was done including Serum lipid profile (LDL of 70mg/dl), ANA by IF, APLA profile and Serum homocysteine level – All of which were normal. 2D Echo and TEE were done with no significant findings.

Therefore initially the cause of Acute Non Hemorrhagic Infarct was considered to be hypercoagulable state due to the intake of oral contraceptive pills and tranexamic acid.

The atypical presentation of stroke in young female patient with bilateral ICA stenosis prompted a DSA to be done which had a characteristic Bilateral complete ICA occlusion with multiple small vessel collateral formation at the basal part with a puff of smoke appearance s/o Moyamoya disease [Figure 3]

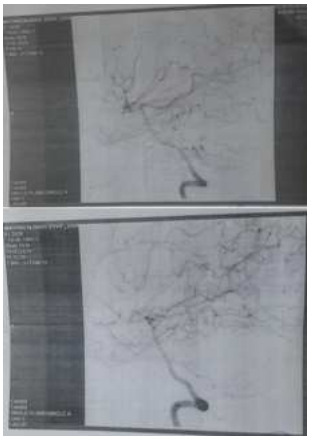

Figure 3 – Moyamoya vessels present with Bilateral ICA occlusion on DSA

The patient was treated with a single antiplatelet and statin with prophylactic dose of anticoagulation. She was also managed for diabetes with Basal bolus regimen and Hypertension with ACE inhibitor. Patient has been advised EC-IC bypass by interventional neurologist however the patient opted for conservative medical management at present.

The patient showed gradual improvement in terms of consciousness. However the left sided hemiparesis showed no recovery. Eventually the patient was discharged on single antiplatelet that is Aspirin 150mg and Atorvastatin 40mg with prophylactic dose of anticoagulation.

DISCUSSION:

Moyamoya disease is one of the infrequent causes of ischemic stroke especially in young individuals. Diagnosing Moyamoya disease is quite challenging and requires knowledge about clinical and radiological features with a high degree of suspicion.

It is an idiopathic disorder with no specific etiology that has been identified. However multiple genetic associations have been recognized with most being linked to ring finger protein 213 gene polymorphisms [4]. Patients with this genetic mutation have a much severe disease with an earlier onset of presentation. It is called Moyamoya disease when the cause is unknown or due to a genetic susceptibility whereas, the term Moyamoya syndrome is used when the vascular findings are due to an associated condition such as tumours, meningitis, haematological conditions like thalassemia or sickle cell disease, vasculitis and other autoimmune disorders, numerous genetic and developmental disorders such as Down's or Marfan's. In this case, the patient has no developmental disorders and the entire thrombophilia screen was negative [5].

Pathologically Moyamoya disease is characterised by hyperplasia of tunica intima with waving of internal elastic lamina of affected vessels. Hemorrhage is cause due to thinning of the tunica media resulting in pseudoaneurysms [6]. Most commonly it affects the anterior circulation however 50 % cases with posterior circulation affliction have also been reported.

Moyamoya disease has a varying neurologic deficits ranging from ischemic stroke and recurrent Transient Ischemic Attacks (TIA) in children to intracerebral hemorrhage in adults. It can also present with headache, seizures or other hyperkinetic movement disorder like chorea or dystonia. In our patient the presentation was with left side hemiparesis diagnosed as Acute non haemorrhagic infarct in the Right MCA territory.

The diagnosis is made by recognising the characteristic angiographic appearance of bilateral stenosis affecting distal internal carotid arteries or other proximal Circle of Willis vessels along with prominent collateral vessels. Digital Subtraction Angiography is the gold standard while CT and MR angiogram have a high diagnostic value as well. In this case there was bilateral ICA and ACA stenosis with right MCA stenosis on MRI angiography which led to suspecting Moyamoya disease that was diagnosed on the DSA. 'Ivy sign' or 'Brush sign' can also be present on MRI. In recent years, High Resolution – Vessel Wall Imaging (HR-VWI) has emerged as an advanced noninvasive technique utilizing multiple contrasts to enable detailed characterization of vessel walls. This innovation significantly enhances diagnostic accuracy for various cerebral vasculopathies like Moyamoya disease when compared to conventional imaging methods. HR – VWI can also differentiate between the disease and syndrome. Based on angiography, Suzuki staging was developed in 1969 and is still in use today [7].

The leading choice for primary treatment in Moyamoya disease is surgical revascularizsation. Surgery is beneficial either by direct or indirect approach particularly if the diagnosis is made early. There are no definite guidelines on medical management. Medical management is not as beneficial and includes antiplatelet – Aspirin or clopidogrel with seizure prophylaxis. Precautions involve lifelong use of anticoagulants (such as apixaban), antihypertensive medications, diabetes management and statins for cholesterol management, and adopting dietary adjustments to promote a healthy lifestyle [8] .Surgical intervention has been superior as it had the longest interval period between further strokes and it reduced the rate of further cerebral ischemic events (1.9%) compared to the antiplatelet therapy group (5.7%) and the conservative management group (15.1%) in a recent retrospective cohort study [9]. Our patient was advised surgical revascularisation that is EC-IC bypass however she opted for conservative medical management and hence was continue on the same.

Therefore it is important for both the neurologist and radiologist to identify the clinical and imaging features of this condition for early diagnosis and management. This rare case therefore highlights the importance of further investigation in younger patients with a history of strokes.

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