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Case Report

Moyamoya disease-the culprit in a young adult presenting with seizures: a case report

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ABSTRACT

Moyamoya disease (MMD) is a rare, progressive cerebrovascular disorder characterised by narrowing the internal carotid artery (ICA) branches and forming fragile, abnormal collateral vessels. Patients with this condition often present with symptoms of cerebral ischemia or haemorrhage, and the disease typically follows a progressive course. Moyamoya derives its roots from Japanese literature meaning cloud or haze. Here we discuss a 23-year-old male who presented to the emergency with the chief complaint of a new-onset single episode of generalized tonic-clonic seizure. On further evaluation found to have MMD and cerebral revascularisation led to a favourable outcome.

Keywords: Moyamoya disease, Transient ischemic attacks, Internal carotid artery

INTRODUCTION

Moyamoya disease (MMD), 1st identified in Japan in 1957, is a rare cerebrovascular disorder marked by the gradual narrowing of the ICA branches, resulting in the formation of abnormal, fragile collateral vessels. Term "Moyamoya," meaning "puff of smoke" in Japanese, was coined by physicians Suzuki and Takaku in 1969, referring to hazy appearance of these collateral vessels on angiographic images in affected patients. The disease is also observed to have a bimodal age distribution, with incidence peaks around 10 years of age and again between 30 and 40 years.[1] MMD has the highest incidence among Asians, particularly in Japan, with a male-to-female ratio of about 1:1.8. While the steno-occlusive regions in MMD generally affect both sides of the brain, there are documented cases of unilateral involvement.[2]

The most common initial symptom of MMD is an ischemic stroke, with transient ischemic attacks (TIAs) also frequently occurring and potentially recurring. Less commonly, disease may present with headaches/seizures.[3]

Here we report a young male presented with seizures with no focal neurological deficits on further evaluation found to have MMD.

CASE REPORT

A 23-year-old male presented to the emergency department with a history of one episode of new-onset generalized tonic-clonic seizure. The patient regained consciousness 20 minutes after the episode and was rushed to the emergency. There was no history of trauma, fever, headache, or similar episodes in the past. The patient's medical history was non-significant.

On examination, his blood pressure (BP) was 138/86 mmHg, pulse rate (PR) was 102 bpm, and his oxygen saturation was 98% on room air with random blood sugar

(RBS) was 112 mg/dl. He had a Glasgow coma scale (GCS) score of E4V5M6, and his pupils were symmetrical and reactive to light. Higher mental functions were normal, and there were no focal neurological deficits. Babinski on the bilateral limbs was down going, and cerebellar examination was normal. There was no sign of meningeal irritation. No visual disturbances were noted.

Routine blood investigations like complete blood picture, serum electrolytes, renal function tests and liver function tests were done within normal limits. An NCCT brain revealed the presence of an intracerebral haemorrhage (ICH) as shown in (Figure 4 A). A fundus examination ruled out papilledema. Further evaluation with MRI brain and MR angiography revealed marked attenuation of the calibre of the left ICA from the C2 to C7 segment. Multiple small collateral arteries were identified in the bifurcation region of both ICAs, the suprasellar region, surrounding the optic chiasma, and in the putamen.

An aneurysm measuring 3.6×3.6 cm was noted in region of right centrum ovale, adjacent to the body of the lateral ventricle in right frontal lobe. This aneurysm was found to arise from a collateral thalamostriate vessel. Flow was absent in M1 segment of both middle cerebral arteries (MCAs) and in the A1 segment of both anterior cerebral arteries (ACAs) as shown in (Figure 1-3). There was evidence of intraparenchymal haemorrhage with intraventricular extension in white matter of right frontal lobe, adjacent to aneurysm. Clinical presentation and diagnostic studies compatible with diagnosis of MMD.

Initially, the patient was managed with inj. mannitol and anti-seizure medication inj. sodium valproate. For definitive management of MMD, the patient was referred to a neurosurgery centre, where he underwent surgical treatment for direct bypass from the superficial temporal artery (STA) to the middle cerebral artery (MCA) with encephalo-duro-arterio-myo-synangiosis (EDAMS), first on the left side and subsequently on the right side. The patient has remained symptom-free since the procedure.

The postoperative image is shown in Figure 5.

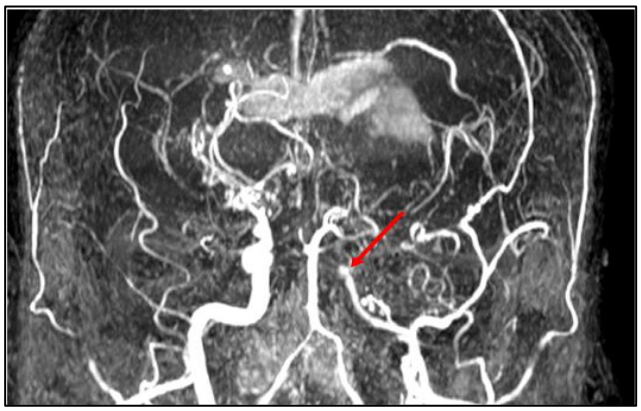

Figure 1: Coronal MIP image showing an attenuated left ICA in calibre (red arrow).

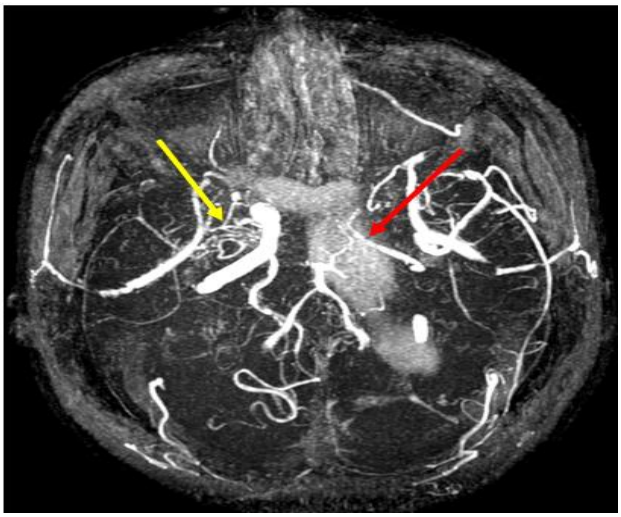

Figure 2: Axial MIP image showing an attenuated left ICA in calibre (red arrow). Multiple collaterals at the site bifurcation of right-sided ICA (yellow arrow).

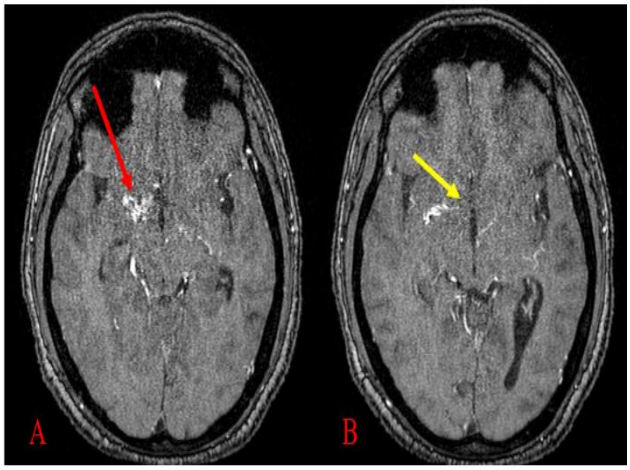

Figure 3 (A and B): Time of flight sequence showing multiple collaterals at the bifurcation of right ICA with absent flow-related signal in A1 segment of bilateral ACA (yellow arrow) and M1 segment of bilateral MCA

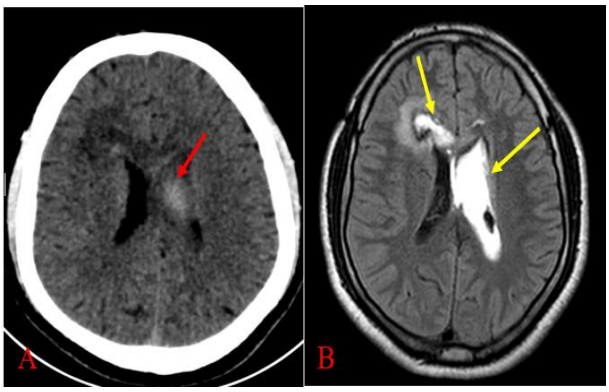

Figure 4 (A and B): Axial NCCT image and T1 axial MRI image showing right intraparenchymal haemorrhage with intraventricular extension.

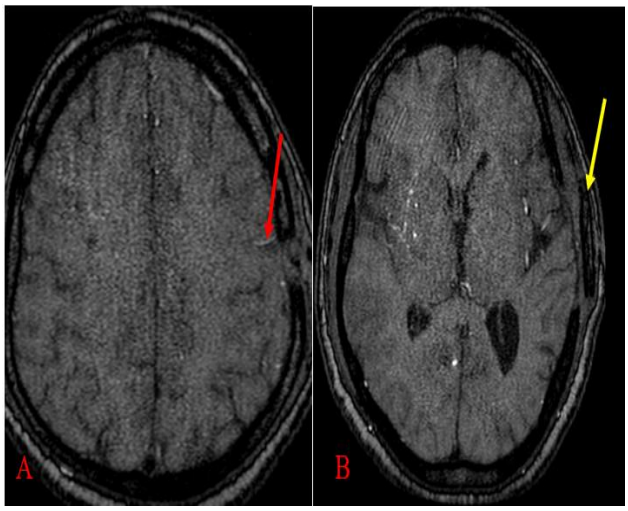

Figure 5 (A and B): A small vascular channel showing a normal flow-related signal originating from the terminal part of the left ACA is seen entering intracranially through a calvarial defect in the left parietal-temporal defect (yellow arrow), extending up to the lateral part of the left pre-central sulcus (red arrow).

DISCUSSION

MMD is a rare and progressive vascular condition where the arteries in the circle of Willis become narrowed or blocked, leading to reduced blood flow to the brain. In response, small blood vessels form at the base of the brain to try to compensate for the reduced circulation.[4]

Diagnosing MMD can be challenging and requires careful consideration of clinical presentation along with appropriate diagnostic imaging. The disease manifests differently across two age groups: in children, typically during the first decade of life, symptoms often include progressive ischemic strokes, TIAs, seizures, and cognitive decline. In contrast, adults, usually between the ages of 30 and 40, are more likely to present with intracranial haemorrhage.[1]

MMD has been linked to various conditions, including immunologic factors like Grave's disease, infectious diseases such as leptospirosis and tuberculosis, and blood disorders like aplastic anaemia, Fanconi anaemia, sickle cell anaemia, and lupus anticoagulant. Additionally, vascular factors such as atherosclerosis, coarctation of the aorta, fibromuscular dysplasia, cranial trauma, radiation injury, parasellar tumors, and hypertension have been associated with the disease. It has also been observed alongside congenital syndromes like Down syndrome, Marfan syndrome, tuberous sclerosis, Turner syndrome, von Recklinghausen disease (neurofibromatosis type 1), and Hirschsprung disease.[2]

MMD can lead to TIAs, strokes, aneurysms, or brain hemorrhages. It may also result in cognitive and developmental delays or disabilities. Cerebral angiography in MMD typically reveals stenosis and occlusion of the arteries in the circle of Willis, along with moyamoya vessels formed as collateral circulation. On MRI, the presence of flow voids in the bilateral basal ganglia suggests moyamoya vasculature. Electroencephalography often shows a reappearance of delta waves after hyperventilation stops, a characteristic finding in MMD.[4] The 90% of MMD patients originally presented with an ischemic stroke, 7.5% with a transient ischemic attack, and 2.5% with a hemorrhagic stroke, according to a worldwide multicenter stroke database.[5]

East Asia has seen the majority of MMD cases, with China, Taiwan, Japan, and Korea having very high annual rates of occurrence. It was demonstrated that this regional and ethnic characteristic is strongly correlated with genetic factors. The polymorphism of R4810K in the RING, which is encoded by the ring finger protein RNF213, which controls protein-protein interactions and possesses ubiquitin-protein ligase activity, was thought to be the most vulnerable gene for Moyamoya illness. Although the pathophysiology of MMD is complex and involves a combination of genetic, environmental, and intrinsic angiogenesis, not all patients with the condition have the RNF213 mutation. The RING finger protein RNF213 is a commonly known susceptibility gene for MMD that has been identified in the family.[6]

In patients with MMD, the basilar artery often shifts toward the midline and moves upward, with posterior circulation vessels appearing enlarged compared to healthy individuals. Disease progression is marked by a longitudinal transition of collateral channels from the anterior to the posterior components, which may elevate the risk of hemorrhagic stroke in adults.[7,8]

MRI not only detects areas of infarction but also provides direct visualization of collateral vessels, seen as multiple small flow voids at the base of the brain and in the basal ganglia. MR angiography is used to confirm the diagnosis and assess the vascular anatomy. It typically shows the narrowing and occlusion of proximal cerebral vessels, along with extensive collateral circulation through perforating vessels, which creates the characteristic "puff of smoke" appearance.[9]

The hallmark of MMD is the presence of bilateral artery angiographic signs of growing anterior circulation stenosis. while unilateral anomalies are the hallmark of Moyamoya syndrome. A primary pathological condition can lead to a secondary pathological process known as Moyamoya syndrome. As an example, patients who receive radiation therapy to the head and neck for cancer may develop significant radiation-induced ICA stenosis in the future. These people eventually develop Moyamoya syndrome, a disorder where the body expands existing smaller blood channels, including the lenticulostriate perforating arteries, in an attempt to compensate for ICA stenosis. Unlike MMD, Moyamoya syndrome often affects just one artery.[10]

Acute management primarily focuses on alleviating symptoms by reducing elevated intracranial pressure, enhancing cerebral blood flow, and controlling seizures.[8] Surgical revascularization has become a promising primary treatment for MMD, especially when medical therapies are insufficient, with documented success. There are two main revascularization techniques used to treat the condition. The direct method involves creating an anastomosis between a branch of the external carotid artery, usually the STA, and a cortical artery. The indirect method places vascularized tissue from the external carotid artery, such as the dura, temporalis muscle, or STA, in contact with the brain. This encourages new blood vessel growth, improving blood flow to the cerebral cortex.[2]

CONCLUSION

This case emphasizes the need to consider MMD as a differential diagnosis for young males presenting with intracranial bleeding. This case highlights the importance that early diagnosis and management lead to favourable outcomes in patients with MMD.

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