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CASE REPORT

'Smoke in the air': a rare cerebrovascular cause of neurological signs and symptoms in a young adult

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Moyamoya disease is a rare neurological condition that affects children and adults of all ages. It is characterized by chronic, progressive stenosis of the circle of Willis that ultimately leads to the development of extensive collateral vessels. Presenting symptoms are usually due to cerebral ischemia or hemorrhage. The Japanese term *moyamoya* (meaning puffy or obscure) was coined to describe the characteristic 'smoke in the air' appearance of these vessels on cerebral angiography. Moyamoya has the highest recorded incidence in Japan (0.28 per 100,000). In the west it is an extremely rare condition with an overall incidence of (0.086 per 100,000) in the Western United States. Etiology for the most part is unknown; however, genetic susceptibility related to RNF213 gene on chromosome 17q25.3 has been suggested. Moyamoya is being diagnosed more frequently in all races with varying clinical manifestations. Moyamoya disease is a rare progressive neurologic condition characterized by occlusion of the cerebral circulation with extensive collaterals recruitment in children and adults. Distinguished radiological findings confirm the diagnosis. Early recognition and swift institution of therapy is vital in order to minimize neurological deficits. We present the case of a 19-year-old African American female who presented with left-sided parastheia, weakness, and headache for 2 days duration.

Keywords: *Moyamoya*; *neurology*; *Japanese*; *circle of Willis*; *collaterals*; *smoke*

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Case report

We present the case of a 19-year-old African American female who presented with left-sided parastheia, weakness, and headache for 2 days duration. Her past medical history was significant for four syncopal episodes lasting less than 30 sec, for which the patient did not seek medical attention. Physical exam revealed decreased light touch and proprioception in the left lower extremity. Laboratory test including complete blood count (CBC), basic metabolic profile, urine drug screen, and thyroid functions were within normal limits.

Computerized tomography of head showed multiple small foci of hypoattenuation in the right periventricular white matter. Contrast magnetic resonance imaging of the brain showed scattered areas of T2 hyper intense signal within the white matter of the right cerebral hemisphere in the 'watershed areas' (Fig. 1). Additional laboratory tests including erythrocyte sedimentation rate (ESR), C-reactive

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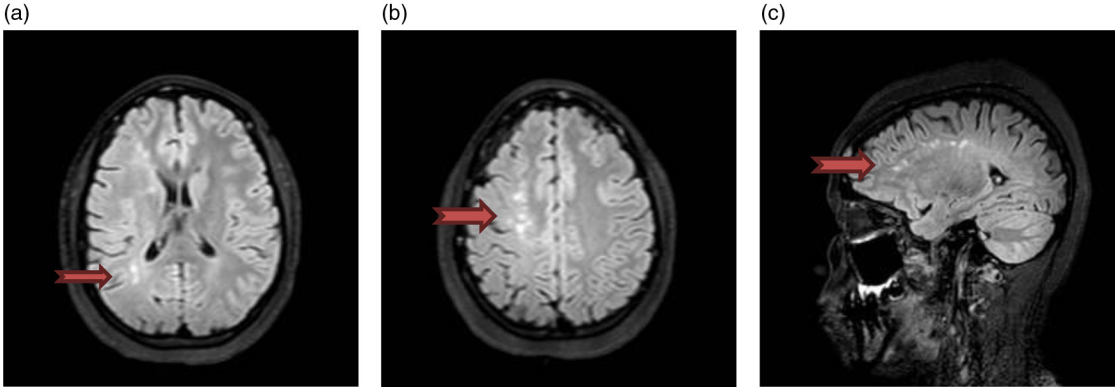

Fig. 1. Contrast magnetic resonance imaging of the brain showed scattered areas of T2 hyper intense signal within the white matter of the right cerebral hemisphere in the 'watershed areas'.

protein (CRP), Antinuclear antibodies (ANA), Cytoplasmic antineutrophil cytoplasmic antibodies (c-ANCA), Perinuclear antineutrophil cytoplasmic antibodies (p-ANCA), protein C and S, antithrombin III, homocysteine, factor V Leiden, coagulation profile, rapid plasma reagin (RPR), HIV, vitamin B12, folate, and a lumbar puncture were all within normal limits.

Magnetic resonance angiography of the brain revealed near occlusion of the distal supraclinoid segment of the right internal carotid artery (ICA) with extensive collateral augmentation (Fig. 2).

Conventional angiography of the head and neck was performed which confirmed the occlusion. Also revealed was a moyamoya-like conversion of the lenticulostriate

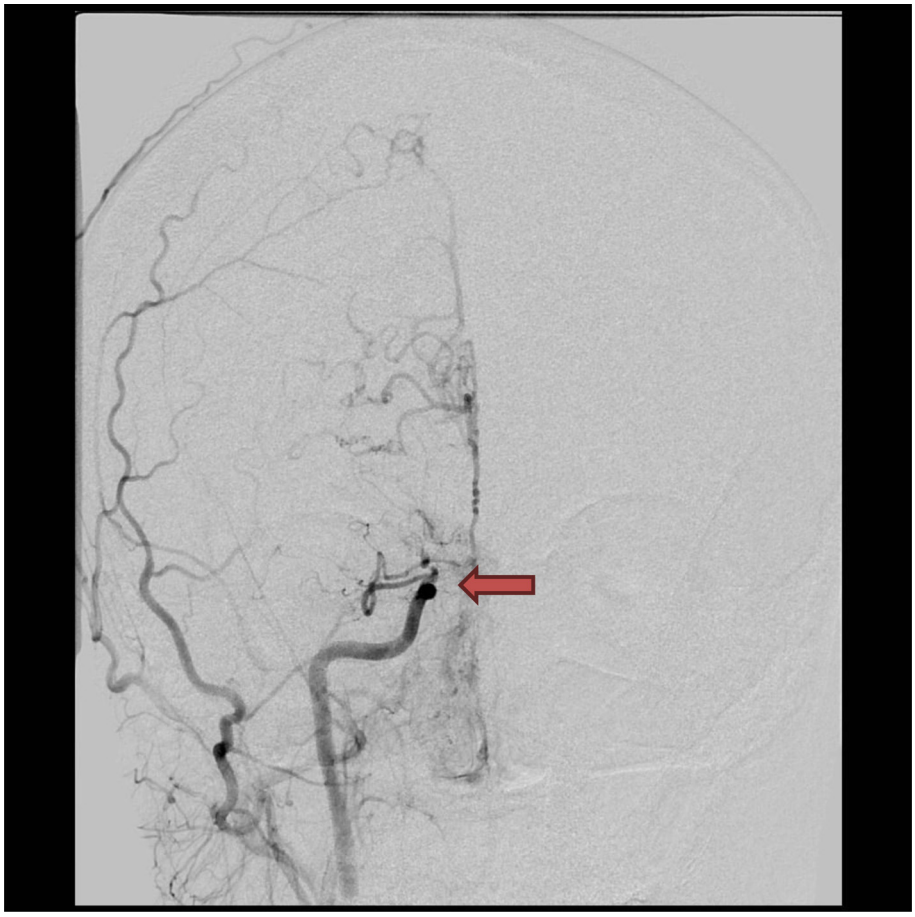

Fig. 2. Conventional angiography of the head and neck showing near occlusion of the distal supraclinoid segment of the right internal carotid artery.

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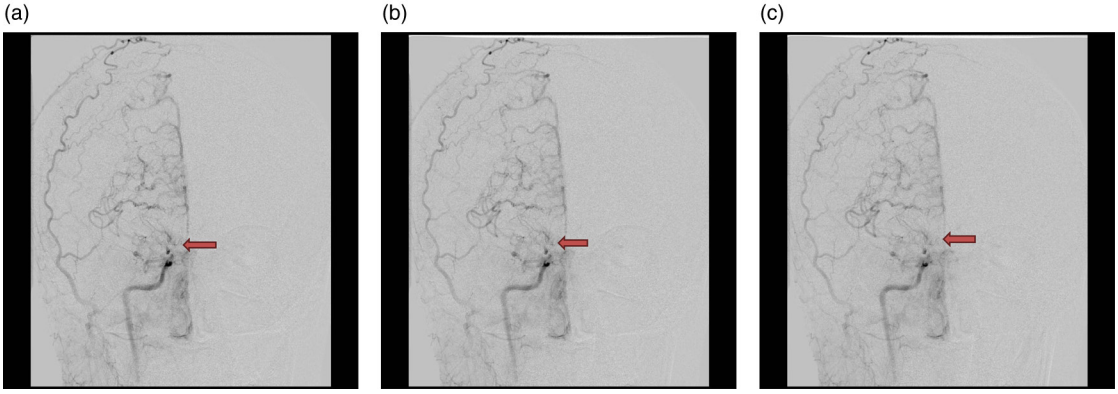

Fig. 3. Conventional angiography of the head and neck showing moyamoya-like conversion of the lenticulostriate arteries giving a classic 'puff of smoke' appearance.

arteries giving a classic 'puff of smoke' appearance (Fig. 3). Based on this, patient was diagnosed with Moyamoya disease. She then underwent neurosurgical anastomosis of the right superficial temporal artery to the right middle cerebral artery.

Discussion

Moyamoya disease (MMD) is a chronic idiopathic neurovascular occlusive disorder that was first described in 1957 by Takeuchi and Shimizu (1). It is characterized by progressive occlusion of the ICA and circle of Willis, leading to formation of collateral small vessels at the base of the brain to compensate the reduced blood flow from arterial stenosis (1). Moyamoya, which means 'puff of smoke' in Japanese, describes the hazy appearance of the abnormal vascular network seen on cerebral angiography (2).

The etiology of MMD is unknown; however, it has been suggested that the onset of the disease can be contributed to genetic, ethnic, and environmental factors (3). There has been a strong link to MMD in Japan and Korea with RNF213, which is a MMD susceptible gene found on chromosome 17q25.3, and mutation p.R4859K (4–6). MMD is more commonly seen in East Asia, particularly Japan, than Europe and the Americas (7, 8). It has been noted that there is a female predominance (9, 10) with a bimodal age distribution involving the first and fourth decade of life (10–13).

The clinical presentation of MMD varies between children and adults. In children, MMD presents with repetitive transient ischemic attacks (TIA) and cerebral infarction. Other symptoms seen in pediatric patients can include episodes of seizures, headaches, strokes, or focal neurological deficits (14). In adults, cerebral hemorrhage, in the intraventricular region, is a common symptom while it is uncommon in children (15). Cerebral ischemia is another presentation seen in adult patients and has been noted to affect the 'watershed' territories of the brain (1).

The main diagnostic imaging studies for MMD include magnetic resonance imaging (MRI), magnetic resonance angiography (MRA), and cerebral angiography. The MRI and MRA are noninvasive imaging studies in MMD that demonstrate steno-occlusive disease of the terminal ICA, the proximal portions of the anterior and middle cerebral arteries, and basal moyamoya vessels, which is the abnormal vascular network in the vicinity of the stenosis or occlusion (16). When the described findings are seen bilaterally on MRI and MRA, it is a definitive diagnosis of MMD and does not require cerebral angiography, while probable diagnosis of MMD is noted if the findings are seen unilaterally (1). Conventional angiography is the gold standard for diagnosis of MMD. It demonstrates fine basal collaterals giving the classic 'puff of smoke' appearance (2). In our case, the MRI showed 'watershed areas' having hyper tense signal within the white matter demonstrating cerebral ischemia, and the conventional angiography of the head and neck in our patient revealed the classic 'puff of smoke' from the conversion of the lenticulostriate arteries.

The mainstay of treatment for MMD is surgical revascularization. Medical treatment has not been proven to be effective (17). Cerebral revascularization techniques include direct and indirect surgical techniques. In the adult population, direct revascularization has proven to be more effective, while in the pediatric patients indirect approach is more successful (1). In adults with MMD having cerebral ischemia, it is recommended that direct arterial bypass with possible indirect surgical techniques is carried out (1).

Conclusion

Moyamoya disease is a rare progressive neurologic condition characterized by occlusion of the cerebral circulation with extensive collaterals recruitment in children and adults. Distinguished radiological findings confirm the diagnosis. Early recognition and swift institution of therapy is vital in order to minimize neurological deficits. This case emphasizes that moyamoya, despite being extremely rare, even more so in African American's, should be

considered in any patient with unexplained neurological symptom indicative of cerebral ischemia.

Conflict of interest and funding

The author has not received any funding or benefits from industry or elsewhere to conduct this study.

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