CASE REPORT

Moyamoya disease

M. Tahir and U. Khan

From the Department of Internal Medicine, Sisters of Charity Hospital, University at Buffalo, Buffalo, NY 14214, USA

Address correspondence to Dr U. Khan. Department of Internal Medicine, Sisters of Charity Hospital, University at Buffalo, 2157 Main Street, Buffalo, NY 14214.
email: usmanayubkhan@gmail.com

Learning point for clinicians
Any patient presenting with significant radiographic findings on CT scan of the head or MRI of the brain that does not correlate with the clinical examination findings should raise the suspicion of secondary pathologies including vasculitides and other rare entities such as moyamoya disease.

Case report

A 33-year-old woman with no significant medical history presented with chronic headache that had been present for the past 2 months. It was reported by the coworkers that she was noted to be more confused. The patient denies any other complaints except for the headache that she localized more to the frontal part of her head. No photophobia, neck pain or fever. Patient denies any sick contacts and recent travel outside the USA, Home medications included pepcid and tylenol. She denied any smoking, alcohol intake and any illicit drug use. She used to work at nail polish saloon.

On examination, vital signs included blood pressure 158/85, pulse 103, respirations 18 and temperature 98.5. Patient noted to be drowsy; however, she is alert and oriented to time, place and person. Patient is also in mild distress due to headache. Head examination was unremarkable. Neck examination was unremarkable with no nuchal rigidity. Extraocular movements were intact, pupils found to be equal, round, reactive to light and accommodation. Chest was clear to auscultation bilaterally, no wheezes or rhonchi. Cardiovascular included normal S1, S2 with regular rate and rhythm; no rubs, murmur or gallops. Abdomen appeared soft, non-tender, non-distended, normal bowel sounds in all four quadrants. Extremities had no cyanosis or edema and no rash or color changes.

On neurological examination, she was oriented to time, place and person. Cranial nerve examination was found to be normal for all, i.e CN 2-12. Motor examination included 5/5 power in all her extremities with normal bulk and reflexes (2+). Sensory examination included no abnormalities. Cerebellar functions were normal, and gait was steady. Kernig’s and Brudzinski's signs were negative.

Laboratory workup included a Complete Blood count with a White blood cells of 7.4, hemoglobin 13.6, platelets 248. Sodium 133, potassium 3.9, Blood urea nitrogen 7, creatinine 0.23, ALT of 13, AST 12, alkaline phosphatase 79. Blood glucose noted to be normal. Urine toxicology was done to rule out as drug intoxication as the cause of the presentation. Urine toxicology was unremarkable.

CT scan head (Figure 1A) showed an impression of areas of infarct predominately involving left temporal and parietal lobes. High density within the areas of infarct likely reflecting cortical sparing.

As depicted in MRI, Figure 1B and 1C shows a large ischaemic infarct involving the left temporoparietal and the left occipital area consistent with infarct of the left Middle Cerebral Artery (MCA) and Posterior Cerebral Artery.

MR angiography (MRA) showed narrowing of the distal ICA bilaterally. The left MCA showed severe attenuation, but the right MCA is less attenuated. The ACAs are also attenuated. The basal artery and vertebral arteries appeared normal.

Cerebral angiogram further strengthened the findings of MRA documenting left P2 occlusion and left PCom occlusion. This angiogram showed a typical appearance of moyamoya disease with severe stenosis to near-occlusions to bilateral terminus ICAs and M1 segments as well as ACAs A1 segments. The left MCA appears to be severely stenotic up to 99% with possible retrograde filling from neovascularization.

Discussion

First described in 1957 as ‘hypoplasia of the bilateral internal carotid arteries’, the characteristic appearance of the associated network of abnormally dilated collateral vessels on angiography was linked to something hazy, like a puff of cigarette smoke, which, in
Japanese, is called moyamoya. It is a unique chronic progressive cerebrovascular disease characterized by bilateral stenosis or occlusion of the arteries around the circle of Willis with prominent arterial collateral circulation. The clinical manifestations of moyamoya are variable and include transient ischemic attack, ischemic stroke, hemorrhagic stroke and epilepsy.

Diagnostic criteria include stenosis or occlusion at the terminal portion of the internal carotid artery, abnormal vascular networks in the basal and presence of these angiographic findings bilaterally. Regarding screening, there is insufficient evidence to screen the asymptomatic individuals or in the relatives of the patients with MMD in the absence of strong family history or medical conditions predisposing to the disease.

Medical treatment only manages the symptoms but not targeted at curing the disease. Several surgical treatments for moyamoya disease have been very effective at bypassing narrowed arteries and creating a new blood supply for the affected areas of the brain, decreasing the likelihood of a stroke. The American Heart Association guidelines note that revascularization surgery is useful for patients who have evidence of low cerebral blood flow or inadequate cerebral perfusion reserve.

The goal of surgical treatment either by direct or indirect technique for moyamoya disease is to reduce the risk of ischemic stroke by improving the cerebral circulation.

The natural history of moyamoya disease tends to be progressive. Untreated patients often suffer cognitive and neurologic decline due to repeated ischemic stroke or hemorrhage.

Conflict of interest: None declared.

References