

Rare disease

Spontaneous occlusion of the circle of Willis in a young woman with epilepsy: epileptic-type moyamoya disease

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The authors report a case of a 31-year-old woman from India with history of seizure disorder who presented with sudden onset right hemiparesis and right-sided upper motor neuron type facial palsy. No identifiable risk factors were noted on admission and all laboratory investigations were negative. MR angiography helped in arriving at the diagnosis of moyamoya disease as the aetiology of her symptoms.

BACKGROUND

Moyamoya disease is a rare cause of ischemic strokes in adults. Seizures in patients with moyamoya disease are mainly described in the paediatric population. The incidence of this disease remains unknown in India. The use of non-invasive imaging modalities should be considered in younger patients presenting with ischaemic symptoms and no identifiable risk factors. This can aid in the diagnosis and early detection of moyamoya disease with possible early intervention.

CASE PRESENTATION

A 31-year-old married woman from New Delhi, India presented with sudden onset right hemiparesis and right-sided upper motor neuron type facial palsy with slurred speech of 6 h duration. She denied having had seizures, headache, trauma, altered sensorium or loss of consciousness prior to the onset of the symptoms. She had no history of cardiac abnormalities, connective tissue disorders, bleeding tendencies, recent infections, cranial irradiation or any risk factors for stroke including hypertension, diabetes mellitus, dyslipidemia, smoking, alcohol or drug use. She denied present or past use of contraceptive pills.

She had a history of seizure disorder for which she had not undergone detailed evaluation and was taking carbamazepine. She had been seizure-free for the past 10 years. She also had a history of hypothyroidism treated with levothyroxine for the past 5 years. Family history was non-contributory.

On examination, she had a pulse rate of 78/min, regular and a blood pressure of 160/90 mm Hg in left arm in supine position. Her body mass index was 22 kg/m². Neurological examination revealed motor aphasia, upper motor neuron type facial palsy on right side and hemiparesis on the right half of the body with the power of 3/5 in upper and lower limbs. Reflexes were brisk on the right side and plantar was extensor. No involvement of other cranial nerves or sensory system was noted. Left side of the body and face were normal. General and other system examinations were within normal limits.

INVESTIGATIONS

Investigations revealed a haemoglobin of 13.3 g/dl, total leucocyte count of 10 900/mm³, platelet count of 394 000/mm³. Liver and renal function tests were normal. Prothrombin time was 10.8 with a control of 10.8

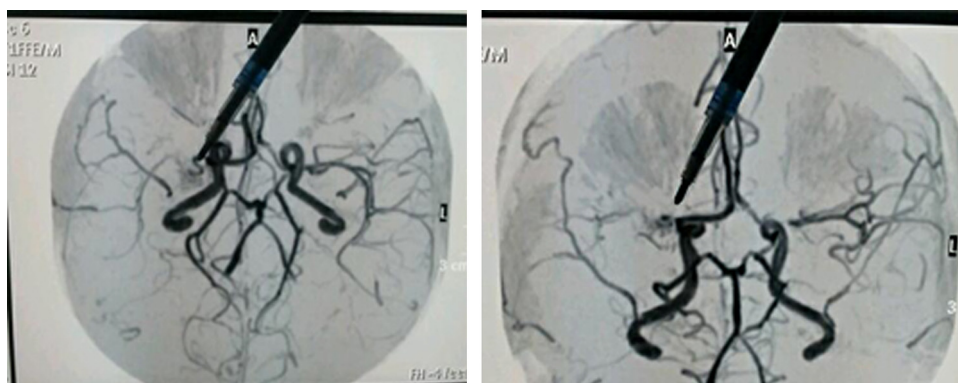


Figure 1 (A, B) MR angiography images of the circle of Willis showing stenosis of right middle cerebral artery (MCA) with multiple collateral formation (see pointer tip). Note the attenuated signal in M1 segment of left MCA and supraclinoid part of left internal carotid artery and the irregularity in A1 segment of left anterior cerebral artery.

(international normalised ratio=1) and activated prothrombin time was 29.2 with a control of 31. Non-contrast CT of the head performed in the emergency department revealed hypodense areas in left periventricular white matter in the temporal and occipital regions with effacement of sulcal spaces consistent with infarction. Electrocardiogram, chest radiograph and 2-dimensional echocardiography did not reveal any abnormality. Carotid Doppler study was unremarkable. She had a triglyceride level of 226 mg/dl and the rest of the lipid profile was within normal limits. Protein C, protein S and factor V Leiden mutation assays were normal. Antinuclear antibody, lupus anticoagulant and anticardiolipin antibody tests were negative. Serum homocysteine level was 6.2 $\mu\text{mol/l}$ (NR 3.3–7.2 $\mu\text{mol/l}$). A MRI of the brain revealed multiple acute infarcts in fronto-parietal region conforming to left middle cerebral artery territory. Subsequent, MR angiography (MRA) (figure 1A,B) showed stenosis of right middle cerebral artery with multiple collateral formations, attenuated signal in M1 segment of left middle cerebral artery and supraclinoid part of left internal carotid artery (ICA) and irregularity in A1 segment of left anterior cerebral artery. MR venogram was normal. This pattern was suggestive of adult-onset moyamoya disease with evidence of bilateral chronic stenosis and formation of collateral vessels on the right side where stenosis was severe. Electroencephalogram done at a later date revealed no abnormality.

DIFFERENTIAL DIAGNOSIS

- 1) Embolic stroke – cardiac versus large vessel related
- 2) Space occupying lesion – tuberculoma, neurocysticercosis, toxoplasma etc
- 3) Vasculitis
- 4) Intracranial haemorrhage – Intraparenchymal, subarachnoid or arterio-venous malformation
- 5) Hypercoagulable states
- 6) Haematological disorders – Sickle cell anaemia, leukaemia's, disseminated intravascular coagulation, polycythemia vera
- 7) Inherited metabolic disorders – Homocystinuria, Fabry's disease.

TREATMENT

She received intravenous hydration and supplemental oxygen. Aspirin (325 mg) was started. Limb physiotherapy and speech therapy were instituted by the third day of admission. Her blood pressure normalised next day without the need for antihypertensive medications.

OUTCOME AND FOLLOW-UP

The patient showed significant improvement in motor power and began to walk with support by the end of first week. She also started to articulate in simple sentences. By the time of discharge from the hospital her power on the affected side was 4/5 in lower limb and 4+/5 in upper limb muscles. The nature and prognosis of the disease were discussed in detail with the patient. She was informed about the medical and surgical interventions available. The

patient declined any surgical intervention at this time and opted for medical management.

DISCUSSION

Moyamoya disease is rare disorder of unknown aetiology characterised by spontaneous progressive stenosis/occlusion of arteries of circle of Willis primarily affecting terminal portion of ICA and their proximal branches namely the anterior cerebral (ACA) and middle cerebral (MCA) arteries. This disease is associated with formation of collaterals near the stenosis appearing as 'puff of smoke' (Moyamoya in Japanese) on angiography.¹

Though more common in Asians, cases have been reported sporadically throughout the world including India.^{2–3} The disease peaks in two age groups: children less than 5 years-of-age presenting with ischaemic strokes and adults in their mid-40s, in whom haemorrhage is often the initial presentation.^{3–4}

Affected patients have symptoms that can be categorised in two main aetiological groups: 1) brain ischaemia (stroke, seizures and transient ischaemic attacks) and 2) consequences secondary to the compensatory mechanisms responding to ischemia (haemorrhage from fragile collaterals, headache from dilated dural collaterals).¹ Seizures although more common in children have also been described in adults in anecdotal reports.⁵

The diagnostic criteria by the research committee on moyamoya disease Japan considers bilateral lesions as definite moyamoya disease.⁶ However, the pathognomonic arteriographic findings in moyamoya disease can vary in severity between the two sides.⁷ These findings may be unilateral initially, however, contralateral disease eventually develops in up to 40% of these patients.^{8–9} Many patients, particularly with ischaemic presentation, are generally not evaluated by angiography and are often missed during initial presentation. Hence, the clinician needs to have a very high index of suspicion to detect this subset of patients.³

There is no treatment available which can reverse the primary disease process. Medical and surgical modalities available aims at improving cerebral blood flow thus protecting against future stroke, reducing collateral formation and reducing the frequency of symptoms. Antiplatelet agents have been used to prevent emboli. Direct revascularisation with anastomosis between branches of internal and external carotid arteries or indirect techniques such as placement of vascularised tissue in direct contact with brain, are the two surgical methods available for these patients.¹

The diagnosis in our patient was made by detection of the lesion by MRA. Formation of collaterals requires a long standing progressive stenosis of the vessels. Our patient's seizure disorder diagnosed 10 years prior could have been an early manifestation of moyamoya disease caused by cerebral ischaemia and perhaps remained undetected as MRA/angiography was not performed at the time of diagnosis. The formation of collaterals as detected on the most recent MRA may have improved the perfusion of the ischemic brain areas and hence explain her symptom-free interval.

Learning points

- ▶ Spontaneous occlusion of the circle of Willis (moyamoya disease) is a rare cause of ischaemic stroke; its incidence is unknown in Indian adults.
- ▶ MRA is an indispensable investigation modality in the evaluation of young patients presenting with ischemic stroke with no identifiable risk factors.
- ▶ Moyamoya disease should be considered as a possible aetiology in the investigation of young patients presenting with neurological symptoms with normal laboratory and routine imaging investigations.
- ▶ Although haemorrhage is the most common initial presentation in adults with moyamoya disease, ischaemic strokes can also be seen.
- ▶ Although bilateral involvement is needed for definite diagnosis, extent of involvement between the sides may vary.
- ▶ Patients presenting with unilateral presentation are likely to eventually develop bilateral disease. Early intervention and treatment might prevent further episodes of neurological complications and should be offered to such patients.

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Competing interests None.

Patient consent Obtained.

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