Moyamoya Disease
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INTRODUCTION
Moyamoya disease is an idiopathic chronic progressive cerebrovascular disease characterized by bilateral occlusion of the arteries around the circle of Willis with prominent collaterals. The age of onset in children is 4-14 years. Arterial ischemic stroke (AIS) is the usual presentation. A characteristic cerebral angiographic appearance is considered the gold standard for diagnosis. Treatment is primarily supportive with close follow-up to assess progression. Antiplatelet therapy is variably used for secondary stroke prevention. Surgical revascularization is indicated for progressive neurological symptoms.

We report the case of a 4 years old boy with acute stroke. This case highlights the fact that noninvasive and less costly MRA can be an invaluable alternative to the conventional cerebral angiography in the evaluation of stroke in children.

CASE REPORT
A 4 years old boy presented to the Emergency Department (ED) with sudden onset of left sided weakness and facial asymmetry. He was completely well until 2 days ago with no antecedent history of fever, flu-like symptoms, headache, visual disturbances or vomiting. Past history of head injury, medication use or blood transfusions was not elicited. The parents reported no cases of early strokes, Ischemic heart disease (IHD), hypertension or autoimmune disease in their family.

On examination, he was a well oriented anxious looking boy with obvious facial asymmetry. There was no dysmorphism and he had normal growth parameters. He was afebrile with a pulse of 92 beats per minute and a respiratory rate of 24 breaths per minute. His blood pressure was 100/60 mmHg. There was no pallor, jaundice or lymphadenopathy. Examination of the motor system revealed grade 0/5 power in the left upper and lower limbs, hypotonia and brisk deep tendon reflexes. The Babinski sign was equivocal. Sensations were intact. There was left sided upper motor neuron type of facial nerve palsy with sparing of the upper part of the face. No other cranial nerve palsies were identified. Fundoscopy yielded normal findings. Systemic review revealed no cutaneous stigmata to suggest a neurocutaneous syndrome, hyperlipidemia or bleeding diathesis. There was no radiofemoral delay, abnormalities on cardiac auscultation, or hepatosplenomegaly.

Investigations revealed Hb of 12.5g/dL. Platelets were 339 x 10^9/L; WBC count was 9.2 x 10^9/L. The blood sugar level was 6.1 mmol/L, ALT value of 28 IU/L and BUN level of 5.2 mmol/L; creatinine was 90 µmol/L with normal serum electrolytes, ESR was 26 mm after first long and a normal lipid profile. His coagulation profile was also normal. Protein C and S concentrations were within normal limits. Antinuclear antibody was negative. Urine for metabolic screening including homocystine levels was normal. An electrocardiogram and echocardiogram revealed no abnormalities. A normal haemoglobin electrophoresis ruled out sickle cell disease.

CT scan with contrast showed multiple areas of non-enhancing hypodensities in the right cerebral hemisphere without mass effect (Figure 1). Magnetic resonance cerebral angiography revealed subacute infarcts in the right fronto-parietal lobe and right basal ganglia. Right internal carotid artery was occluded with distal filling from collaterals (Figure 2). Moyamoya disease was strongly suggested as the probable diagnosis. The patient was booked for cerebral angiography. It further confirmed the diagnosis of Moyamoya disease. Having ruled out hemorrhagic stroke on neuroimaging, the patient was started on oral acetylsalicylic acid. A comprehensive physiotherapy program was instituted. Gradual improvement in muscle strength became noticeable about 2 weeks after presentation.

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DISCUSSION

"Moyamoya" meaning puff of smoke pertains to the smoky angiographic appearance of the vascular collateral network resulting from occlusion of the arteries of the circle of Willis. The primary idiopathic disorder is termed Moyamoya disease, while similar appearance associated with conditions like NF-1, Down syndrome and sickle cell disease is referred to as Moyamoya syndrome. Japanese people are affected more often with a reported annual incidence of one per 100,000.1 Sporadic cases of Moyamoya disease have appeared in Pakistani literature from time to time,2-5 although its true incidence remains undetermined. Familial, genetic and environmental factors are often implicated in this otherwise idiopathic disorder. An association with class II genes of the human leukocyte antigen (HLA) is suggestive of a genetic influence in its pathogenesis. The peak age of onset of Moyamoya disease in children is 4 to 14 years. Another peak occurs in adults between 40 and 50 years of age. Females are affected twice as often as males.

Moyamoya can present as a single episode or multiple recurrences. Affected children usually present with transient ischemic attack (TIA’s) or ischemic stroke. Stroke in children is often a result of Tuberculous meningitis (TBM) and bacterial meningitis.6 The associated features help differentiate these disorders from the stroke in Moyamoya disease which occurs as an isolated ischemic event. Adults with Moyamoya disease usually present with hemorrhagic stroke and occasionally epilepsy.7 The involvement is unilateral in 15% of cases.8 In children symptomatic episodes of ischemia may be triggered by exercise, crying, coughing, straining, fever or hyperventilation. Magnetic resonance angiography (MRA) has replaced conventional angiography in most centers due to its high diagnostic yield and noninvasive nature.9 In fact the availability of MRA is a major development in the evaluation of childhood stroke and is likely to improve Moyamoya disease case detection. This is particularly applicable in countries like Pakistan where conventional cerebral angiography is either unavailable or unacceptable to parents due to its invasive nature and cost.

For patients with Moyamoya disease and acute stroke, aims of treatment initially include reduction of elevated intracranial pressure, improving cerebral blood flow and controlling seizures. Thrombolytic and antithrombotic therapy is not indicated in the acute phase of illness. For Moyamoya syndrome, it is important to search for and treat the underlying condition. A prime example is sickle cell disease in which transfusion therapy is effective for primary and secondary stroke prevention. For asymptomatic or mildly symptomatic Moyamoya disease, acetylsalicylic acid in antiplatelet dose (2-5 mg/kg daily) is indicated as initial therapy. For patients with progressive ischemic symptoms surgical revascularization is the recommended treatment.10 Rehabilitation and physiotherapy for neurological deficit is employed early in the course of the disease to prevent long-term morbidity.

Moyamoya disease tends to be progressive in nature. Close follow-up is essential to recognize and treat repeated ischemic events.

REFERENCES


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