Case Report

Moyamoya disease with rapidly progressive outcome in a child : a case report

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We report a case of 5-year-old female child who was admitted at our hospital with complaints of quadriparesis with aphasia. On MR angiography, the child was diagnosed to have Moyamoya disease. It has slow progression but in our case the clinical progression from asymptomatic to quadriparesis along with cognitive decline and aphasia was rapid over period of 6 months. The patient was treated conservatively and referred to a higher centre for specific neurosurgical intervention.

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***Key* words : Moyamoya disease, quadriparesis with aphasia, cyanotic heart disease.**

Moyamoya disease is a rare idiopathic non-atherosclerotic cerebrovascular disease characterized by stenosis and progressive occlusion of the bilateral distal end of internal carotid artery and its proximal branches. Upon cerebral angiography, this abnormality is typically visualized as a puff of smoke-like pattern due to formation of new collateral vessels at the base of brain. This disorder is progressive in children and relatively stables in adult's. We report this case due to unusual and rapid progression of the disease due to associated risk factors.

Case Report

A 5 years old female child presented with history of weakness of all four limbs since last 6 months, abnormal movements of all four limbs with frothing from mouth 3 month back and inability to speak since last 1 month. She was apparently well till 6 months back when mother noticed paucity of movements and weakness of right upper and lower limb. It was sudden in onset and resolved over period of 1 week but with some residual weakness. A week later she developed sudden onset weakness of left upper and lower limb that is persisting till date. She had single episode of generalized tonic clonic movements 3 months later. Since last 1 month she has developed loss of speech, change in behaviour and drooling of saliva. There was no history of trauma, fever, headache, vomiting, ear discharge, rash, joint pain, repeated blood transfusion, cyanosis, suck-rest-suck cycle, palpitations or breathlessness. There was no history of blurring of vision, deviation of angle of mouth while eating, any involuntary movements, flushing, excessive sweating, bedsores or contractures. There was history of similar episode of weakness of both upper and lower limbs of left side, recovered within a day occurring 1 year back and she was asymptomatic for following 6 months. Birth history was uneventful. Development of child was normal till 6 months back. Family history of stroke in grandmother and maternal uncle was present (Death at 26 years of age). There was no history of tuberculosis in family.

***Examinations* —** On examination child's vitals were stable, she was alert and arousable, Blood pressure in all four limbs were normal, Head to toe examination was normal, no dysmorphic features, no neurocutaneous marker. Neurological Examination reveals that cranial nerves were normal, Gag reflex was present, Aphasia was present, tone increased bilaterally all four limbs, power 3/5 all the limbs (right > left), reflexes were brisk bilaterally, sustained ankle clonus was present and no bladder or bowel involvement. Rest of the systemic examination was normal.

She was diagnosed to have Quadriparesis with aphasia with no cranial nerve or autonomic involvement. She was investigated for hematological diseases (sickle cell disease, hypercoaguable diseases), vasculitis associated with collagen vascular diseases (SLE, RA, scleroderma), vasculopathies (Moyamoya disease, MELAS), cardiac diseases (cyanotic heart disease). Her routine investigation (Table 1) was normal. Chest x-ray, ECG and 2D ECHO were normal. Her Homocysteine levels, serum lactate, C3 and C4 levels were normal and ANA was negative. MRI head was done which revealed large infarct in right frontoparietal region and also infarcts in basal ganglia and thalamus. MR angiography (Fig 1) was done which revealed narrowing of bilateral distal part of ICA and M1 and A1 part of right middle and anterior cerebral arteries respectively. In view of history of recurrent stroke, cognitive and neurological decline, large multifocal infarcts involving areas of major cerebral arteries and characteristic MR angiography findings child was diagnosed of having Moyamoya disease. Child was managed conservatively on Aspirin (Class IIb) and was referred to higher centre for revascularization surgery.

Discussion

Moyamoya disease is a cerebrovascular disease characterized by slowly progressive steno-occlusive changes in the terminal portions of the bilateral internal carotid arteries (ICA) and their main branches, which results in the formation of a fine vascular network at the base of the brain (moyamoya vessels) to compensate for the steno-occlusion[1]. The hazy appearance of these hypertrophied collaterals on angiography resembles a puff of smoke (moyamoya in Japanese); thus, Suzuki and Takaku named this novel disorder "moyamoya disease"[2]. In 1997, the research committee headed by Fukui M[3] published guidelines for the diagnosis of moyamoya disease. Definite moyamoya disease is diagnosed when conventional angiography shows the following findings :- Stenosis or occlusion

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Table 1 — *Laboratory parameters*

Blood tests	At Presentation
Hemoglobin, g/dL	9.1
White blood cells, cells/mm^3	11,500
Platelets, cells/mm^3	6.5
Sodium, meq/L	138
Potassium, meq/L	3.6
Calcium, mg/dL	9.6
Phosphorus, mg/dL	4.5
Blood urea, mg/dL	23
Serum creatinine, mg/dL	0.6
Uric acid, mg/dL	4.0
Glucose, mg/dL	100
Albumin, g/dL	3.7
Homocysteine levels	6.35 (4.44-13.56)
Lipid Profile	normal
C3	138 mg/dl (80-160 mg/dl)
C4	32.5mg/dl (20-40 mg/dl)
ANA	negative
Serum Lactate levels	1.5 mmol/L (0.7-2.1)

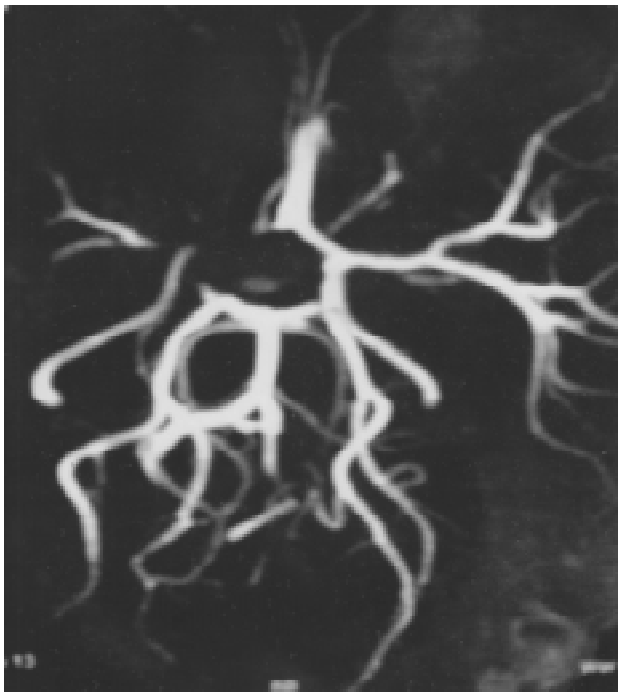

Fig 1 — Showing MR angiography – Bilateral ICA narrowing and narrowing of M1 and A1 part of Right MCA and ACA

in the terminal ICA and/or proximal portion of anterior cerebral artery (ACA) and/or middle cerebral artery (MCA) and abnormal vascular networks (moyamoya vessels) in the basal ganglia and bilateral lesions. This classification was based on cerebral angiography but in children MR angiography findings as in our case were also included for definite diagnosis of Moyamoya disease[4].

It is a rare disorder. In Japan it is most common pediatric cerebrovascular disease affecting girls twice as often as boys[5]. Incidence and Prevalence in Japan 0.94 and 10.5/100,000[5]. It affects American Asian more than Blacks than Hispanics[6]. Peak incidence is first decade of life and 30-40 years[6]. Familial Incidence of affected first-degree relatives is 7%-12% in Japan[5]. Incidence in Indian children is unknown. Our case has presented early at 4 years of age.

The natural history of moyamoya disease tends to be progressive. Children often suffer cognitive and neurologic decline due to repeated ischemic stroke or hemorrhage which was seen in our case. Moyamoya may have a more rapid progression and a worse prognosis in younger than in older children[7]. According to study by Ishii *et al*, childhood patients with MMD progressed within 5-10 years to more severe stages angiographically while some of the cases progressed after adolescence[8]. In our case the clinical progression from asymptomatic to quadriparesis along with cognitive decline and aphasia was rapid over period of 6 months which could be due to younger age at onset and possible genetic association as was evident in family history. The researchers suggest predictor of progression as follows younger age at onset (<7 years), family history of MMD, asian heritage, cranial irradiation and previous history of cardiac abnormalities[9]. Acute management is mainly symptomatic and directed towards reducing elevated intracranial pressure, improving cerebral blood flow, and controlling seizures. Revascularization procedures are currently performed to increase the perfusion to the hypoxic brain tissue[10].

Moyamoya disease is an important cause of cerebral stroke in children, especially in east Asian countries. For the pediatrician it is important to be familiar with the clinical manifestations and MRI/MRA findings in moyamoya disease or syndrome to make an early diagnosis leading to a good prognosis. Genetic analysis of familial moyamoya disease might help to determine the pathogenesis in the near future. Role of screening is in those with recurrent stroke with incomplete recovery ,large multifocal infarcts in MCA /ACA, strong family history of moyamoya and medical conditions associated with moyamoya syndrome.

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