

CASE REPORT**Moya Moya Disease: Presenting With Changing Neurological Signs**Gurudutt Joshi¹*¹Department of Pediatrics, Surat Municipal Institute of Medical Education and Research (S.M.I.M.E.R.), Sahara Gate, Surat - 395001(Gujarat), India**Abstract:**

A 4 yrs old female child was admitted with left hemiparesis and unconsciousness and a short history of neurological impairment, changing neurological signs with progress of the disease and most of relevant laboratory investigations being normal, child underwent MRI and MRA which revealed features of Moya moya disease.

Keywords: Moyamoya disease, puff of smoke, MRI.

Introduction:

Moyamoya is a rare idiopathic progressive vaso-occlusive disease characterized by irreversible occlusion of main blood vessels to the brain as they enter into the skull. The occlusive process stimulates the development of an extensive network of enlarged basal transcortical and transdural collateral vessels [1, 2]. Kudo first reported this pattern of collateral flow in 1960 [3]. This process may be due to various external stimuli, injuries, hereditary defects but in some cases no etiology can be found, as a result patients may present with various neurological signs and symptoms which may be progressive and changing.

Case Report:

A 4 years old female child presented with inability to walk since one week, unable to speak, eat sit, deviation of mouth and unconsciousness, since one day. On admission, she was diagnosed initially as left hemiparesis, ipsilateral facial palsy, generalized hypotonia, coma but no signs of meningeal irritation, later unilateral hemiparesis developed into quadriplegia within 2-3 days. She had a history of generalized tonic clonic convulsions and unconsciousness before 8 months however no investigations were carried out at that time and another episode of history of difficulty in walking

and weakness of left lower limb before 2 months of admission, from which she recovered within one week. There were no clinical or radiological evidence of T.B. Her routine immunization was complete for the age except the last boosters for OPV and DPT. Her developmental at history was normal and there was no history of exposure to cranial irradiation. On clinical examinations the other systems were normal. Her laboratory investigations showed normal complete blood count serum electrolytes, cerebro spinal fluid coagulation and lipid profile, renal function tests and liver function tests. Test for sickling, M.T. and P.S. M.P. were negative. Chest X-ray and fundus were also normal.

Her initial MRI report showed areas (Acute infarcts) involving the right fronto-parietal region (Fig. 1) and subsequent MRI after a week showed multiple sub-acute infarcts/ischemia in right fronto-parietal lobes in right.

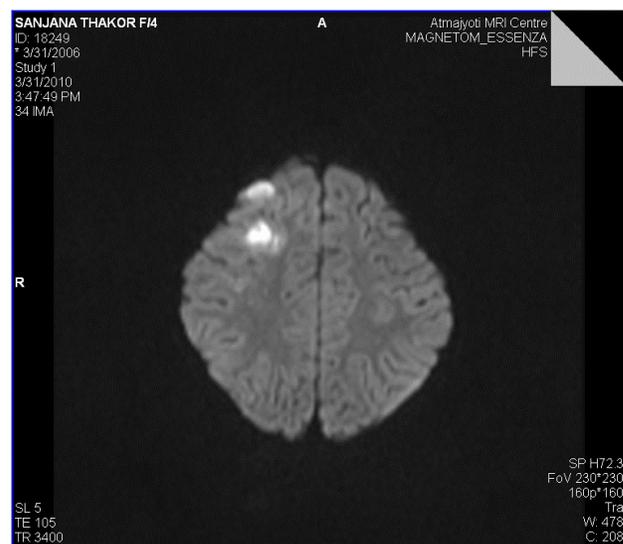


Fig. 1: Infarct Right Fronto-Parietal Region

MCA territory area: Similar multiple small recent infarcts/ischemia is also detected in left fronto parietal, temporal and left basal ganglia in left MCA territory (Fig. 2)

MR angiography images through circle of willis revealed progressive narrowing in supraclenoid

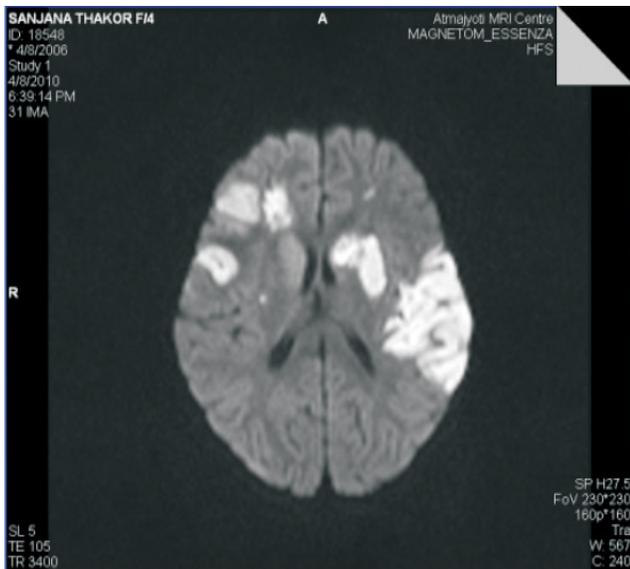


Fig. 2: Multiple Infarcts Right and Left Fronto-Parietal, Temporal, and Left Basal Ganglia.

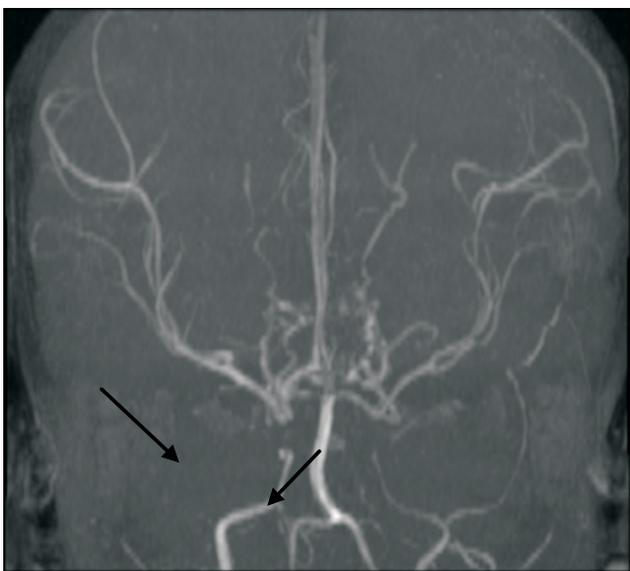


Fig. 3: Narrowing in Supraclenoid Portion of both ICA

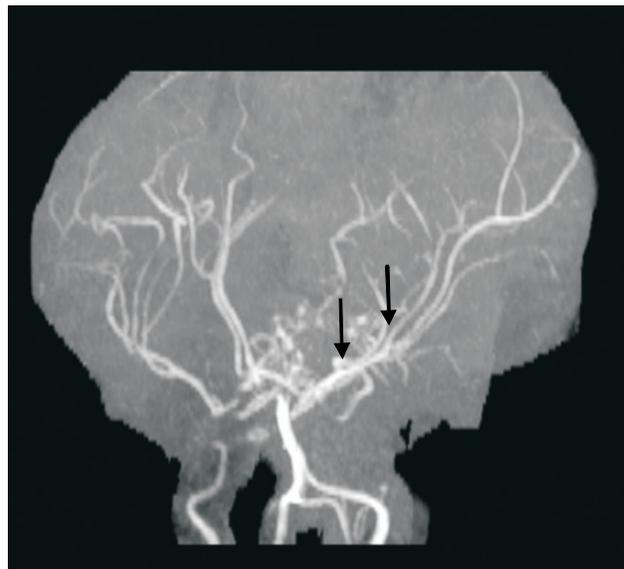


Fig. 4: Few Thalamoperforators (Moyamoya) Collaterals in Peri-Mesencephalic Cistern

portion of both ICA. Subtle narrowing is also seen in cavernous and petrous portion of left ICA. (Fig. 3) Few thalamo perforator collaterals (Moya Moya collateral) detected in ambient and peri- mesencephalic cistern, gives “Puff of smoke” appearance. (Fig. 4)

Discussion:

The word *moya* is Japanese, and is taken to mean “tiny”, referring to the tiny vessel network that forms. Moyamoya disease although reported in Japan, affects all races. Precise cause of the disease is unknown [4]. It is a chronic, progressive, non-inflammatory vasculopathy resulting in slowly progressive, bilateral occlusion of the internal carotid arteries starting at carotid syphon.

The basilar artery is sometimes occluded as well, because the occlusion is slowly progressive multiple anastomoses form between the external and internal carotids. The result is new vascular network at the base of brain composed of collaterals from anterior or posterior choroidal arteries. On MR angiography, these telangiectasias produce a hazy appearance, like “Puff of Smoke” [5]. In children, Moyamoya disease presents with symptoms suggestive of ischemia of brain

with episodes of seizures, focal neurological deficits, recurrent strokes or alternating hemiplegia. It is associated with Sturge weber syndrome, neurofibromatosis, Down syndrome, Fanconi's anemia, sickle cell anemia, cranial irradiation, genetic [4].

It is characterized by stenosis or occlusion of terminal portions of the intracranial internal carotid arteries and the proximal portions of the anterior and middle cerebral arteries with abnormal vascular networks in their vicinity visible as "puff of smoke" on MR angiography.

MRI with angiography is the investigation of choice for moya moya syndrome [6]. Findings of this nature bilaterally are considered definitive of Moyamoya disease if unilateral as "probable cases" [7-10]. According to the above guidelines our case has MRI and MRA findings similar to the definite of Moyamoya disease. However there was a history of fall in the past with generalized tonic and clonic convulsions and unconsciousness which was either due to head trauma per se or because of the cerebro-vascular blockage could not be concluded as she was not investigated at that time.

Around 50% of children with Moyamoya disease will have no long term sequel if left untreated [4]. This

patient presented with initial facial paralysis, hemiparesis, progressing to quadriplegia from which she had partially recovered at the time of discharge and recovery is still progressive on follow up. Most of the relevant investigations were normal. Provisional diagnosis of vaso-occlusive disease with such presentation may have to be kept in differentiating the conditions associated with changing neurological patterns and of sudden onset and radiologically investigated with neuroimaging.

Synangiosis is the treatment of choice, with some variations in the origin of the vascular supply of graft which are useful to confirm the diagnosis is MRI, MRA and the operations described are
EDAS: Encephaloduroarteriosynangiosis
EDMS: Encephaloduro arteriomyosynangiosis
EMS: Encephalomyosynangiosis [11]
STA: MCA superficial temporal to middle cerebral artery anastomosis.

Conclusion:

Any child presenting with changing neurological signs with convulsions should be considered to have rare conditions like Moya moya disease in differential diagnosis, especially when routine CNS investigations are normal.

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