Unravelling Paediatric Stroke: A Rare Presentation of Paediatric Moya Moya Disease with Simultaneous Involvement of Anterior and Posterior Circulation

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Abstract

Moya Moya disease is a rare cerebrovascular disorder characterized by progressive stenosis of the internal carotid arteries and their branches, leading to a range of neurological symptoms. This case report discusses a 14-year-old boy presenting with classic symptoms associated with Moya Moya disease. The diagnostic process involved imaging studies, which confirmed the characteristic findings of the disease along with a specific ANA study which further helped in excluding the association of any other autoimmune disease. Treatment options were explored, including, the use of therapeutic drugs like Aspirin and some statins, aimed at preventing TIAs and alleviating symptoms. This case highlights the importance of early recognition and management of Moya Moya disease to prevent serious neurological deficits and improve the quality of life in affected children and adolescents. Further research is needed to explore the long-term outcomes of various treatment strategies for this challenging condition.

Keywords: *Moyamoya, Paediatric, Angiography*

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Introduction

Moyamoya disease is a rare, progressive cerebrovascular disorder of unknown etiology, characterized by stenosis or occlusion of the arteries in the Circle of Willis and the subsequent formation of compensatory collateral vessels, which appear as a "puff of smoke" on cerebral angiography (1). The condition is most prevalent in East Asian populations and exhibits a bimodal age distribution, affecting children around 5 years and adults around 40 years. Its pathogenesis is thought to involve genetic predispositions, particularly mutations in the RNF213 gene, as well as autoimmune and environmental factors. Clinical presentation typically includes transient ischemic attacks, seizures, headaches, visual disturbances, and cognitive decline. Diagnosis is predominantly radiological and done with neuroimaging modalities. A related entity, Moyamoya syndrome, presents with similar angiographic findings secondary to other conditions, including Down syndrome, hyperthyroidism, and neurofibromatosis type 1 (3). Although there is no definitive preventive strategy, treatment includes antiplatelet agents, anticonvulsants, surgical revascularization, and rehabilitative support.

Case Report

A 14-year-old male was brought to the Emergency Department at North Bengal Medical College and Hospital, West Bengal, India, with a history of recurrent episodes of transient weakness in the left upper and lower limbs over the past eight months. Each episode had a sudden onset and typically resolved within one to two hours. During these episodes, the child experienced difficulty in standing or walking and often fell,

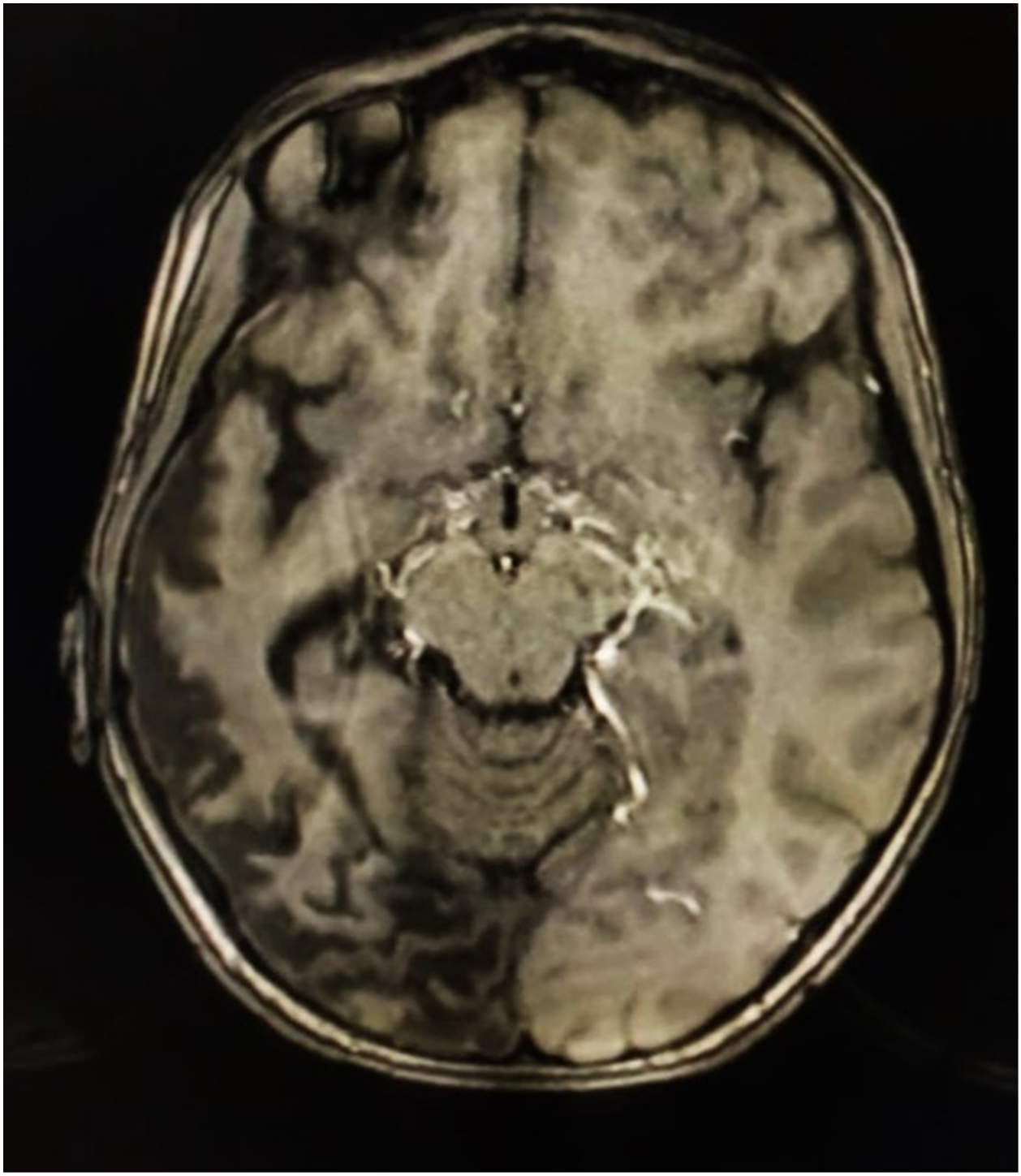

Figure 1: Diffuse atrophy in right MCA and PCA territory with multiple tiny collaterals on TOF MRA. Major vessels of the Circle of Willis not visualised.

requiring assistance to get up. The events were not associated with loss of consciousness, seizures, vomiting, tongue biting, or bladder or bowel incontinence. There was no history of visual disturbances, dizziness, headaches, or convulsions.

He was the first child of non-consanguineous parents and was born via normal vaginal delivery in a hospital setting. Upon admission, a preliminary physical examination was conducted. The child's vital signs were within normal limits: blood pressure was 100/60 mmHg, and pulse rate was 70 beats per minute. Anthropometric parameters were appropriate for age. Neurological examination showed normal muscle tone and strength (5/5) in all limbs. Deep tendon reflexes, including ankle and knee jerks, were

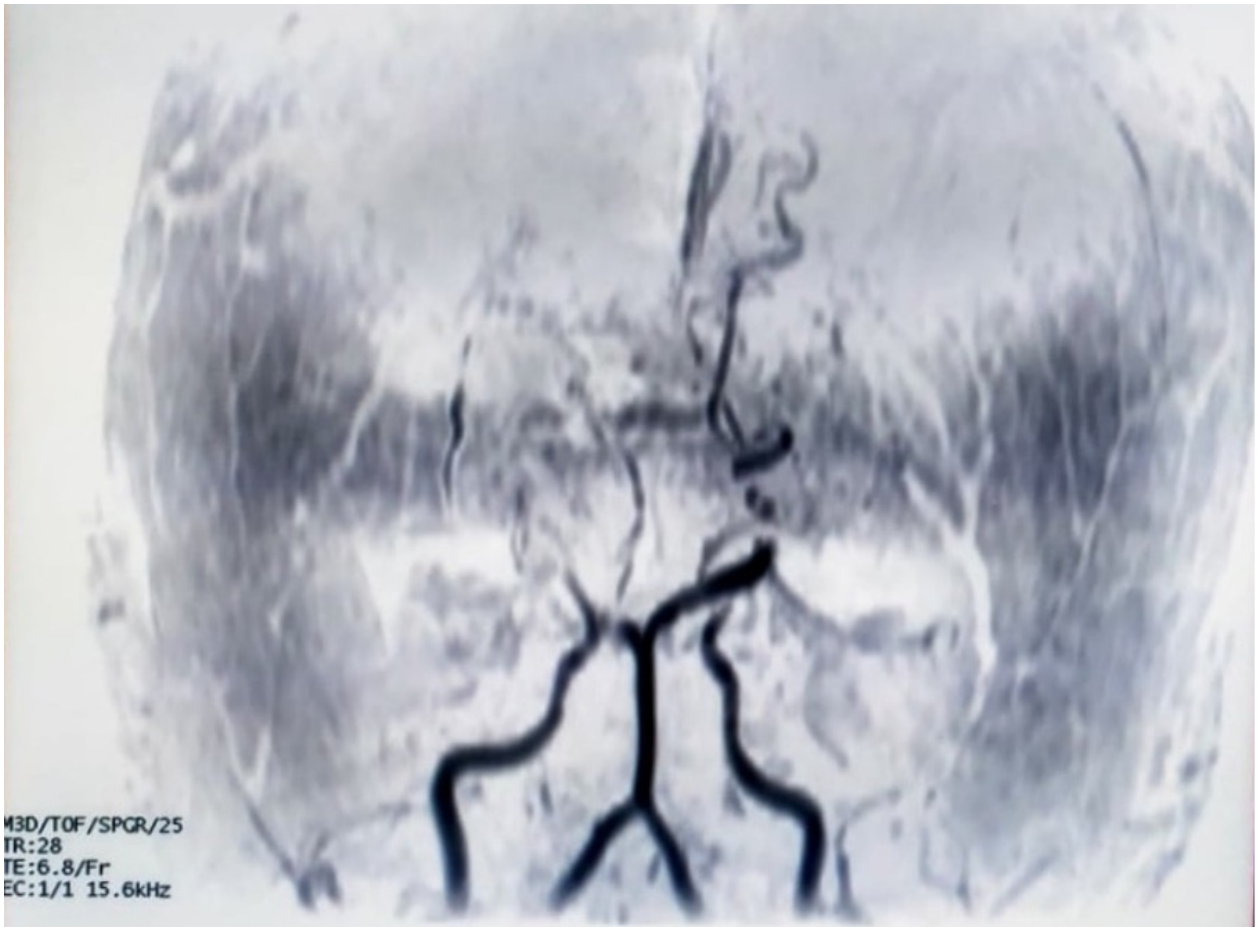

Figure 2: 3D MIP TOF MRA coronal image showing the absence of supra clinoid ICA, MCA, and PCA with multiple serpentine vessels giving a "Puff of Smoke" appearance.

brisk bilaterally. The plantar reflex was flexor on the right and absent on the left. No significant sensory deficits or abnormalities were detected on systemic examination. Immediately after the preliminary examination, the child was sent for radiological imaging, as per the stroke protocol of the institution. MRI revealed diffuse parieto-occipital atrophy predominantly in the right Middle Cerebral Artery (MCA) and Posterior Cerebral Artery (PCA). No evidence of restricted diffusion was seen. A gyriform high signal was seen in the posterior parietal and occipital convexity on Fluid-Attenuated Inversion Recovery (FLAIR) images, likely indicating sequelae of previous infarction. TOF-MRA (Time of Flight Magnetic Resonance Angiography), revealed occlusion of the terminal ICAs and their proximal branches, with the formation of extensive collateral networks, indeed confirming the diagnosis. There is non-visualization of both internal carotid arteries (ICA) beyond the supra-clinoid segments. On the left side, neither the supra clinoid segment of the ICA, nor the anterior cerebral artery (ACA), nor the MCA were visualized, indicating severe occlusive disease. On the right side, a small segment of the terminal ICA was visualized, which gave rise separately to both the right and left ACA. Additionally, a short segment of the right M1 (MCA) is seen. The left PCA was visualized up to the P4 segment; however, the vessel demonstrates reduced caliber, suggestive of hypoplasia or diminished flow. The right PCA was not visualized beyond the P1 segment. The right posterior communicating artery (PCoM) was not visualized. Multiple tiny serpiginous collateral vessels are noted in the expected locations of the absent major vessels and the basal ganglia, forming a network of abnormal vascular channels **(Figure 1)**. These collaterals appeared as the classic "puff of smoke" appearance on 3D maximum intensity projection MRA TOF images **(Figure 2)**. A provisional diagnosis of Moyamoya disease was made.

The child was then kept under observation and monitored for any further signs and

symptoms. The child was started on Aspirin to prevent the recurrence of transient ischaemic attacks along with some Statins. The blood sample was also sent for Anti-Nuclear Antibody (ANA) by Immunofluorescence (IF) and blot on the 3rd day of ad-mission, to rule out the underlying presence of any other autoimmune disease, and Nucleoli 2+ pattern by IF was observed, and ANA immunoblot report was found to be negative.

The child's sensory and motor systems were closely monitored during hospitalization, with no further deterioration observed. He remained clinically stable on basic medical management. After discussing therapeutic options, the parents opted for conservative treatment. The child was discharged after five days of observation. At the time of discharge, his general and systemic examinations were normal, with no significant changes in motor function. He was advised to attend regular follow-up visits for ongoing evaluation. The family was counseled to avoid situations that might trigger hyperventilation, crying, or straining, as such conditions could reduce carbon dioxide levels and precipitate transient ischaemic attacks.

Discussion

Moyamoya disease is a progressive cerebrovascular disorder characterized by occlusion of the internal carotid arteries (ICAs), primarily due to intimal thickening. This leads to the development of collateral vessels around the Circle of Willis, producing the classic “puff of smoke” appearance seen in angiography (2). Although typically bilateral, unilateral involvement is also reported. The disease shows a higher prevalence in East Asia, especially in Japan and China, and occurs more commonly in females than males (1:1.8). The age of onset is generally between 10–14 years in males and 20–24 years in females.

The clinical presentation varies by age. In children, symptoms often include transient ischemic attacks (TIAs), seizures, cognitive impairment, speech difficulties, and body weakness (7). In adults, strokes, headaches, and speech disturbances are more common. Our 14-year-old male patient presented with repeated transient episodes of left-sided limb weakness over 8 months, without loss of consciousness, seizures, or incontinence. On examination, tone, and power were normal in all four limbs, with brisk knee and ankle reflexes and an absent left plantar response. No sensory deficits were noted, but cognitive decline and poor scholastic performance were present. Our case was unique in the aspect that both the anterior and posterior circulation were simultaneously involved in the disease process which is not a frequent finding in this disease.

Moyamoya disease has been associated with genetic and environmental factors. A strong link exists with mutations in the RNF213 gene on chromosome 17. Inflammatory and angiogenic mediators such as VEGF, PDGF, TGF-β, and HGF are also believed to play a role (5). To investigate potential immune involvement, ANA testing was done. A Nucleoli 2+ pattern was observed on immunofluorescence (IF), but the ANA immunoblot was negative. While some cases report positive ANA titers in Moyamoya syndrome with concomitant autoimmune conditions like SLE, such an association was not found in our case (4).

Children with Moyamoya are sensitive to triggers such as crying, hyperventilation, or straining, which lower carbon dioxide levels and can provoke TIAs (8). The child’s family was advised to avoid such stimuli. Treatment includes long-term antiplatelet therapy, such as aspirin and cilostazol, to prevent recurrent TIAs (6, 9). Revascularization surgery is another option to restore cerebral perfusion, though it does not reverse the disease process. It significantly reduces the risk of future strokes but cannot eliminate it (9).

Conclusion

Moyamoya disease is a vasculopathy of unknown origin and a common cause of pediatric stroke. Clinicians must be aware of this rare entity while evaluating cases of pediatric strokes. Neuroimaging with angiography is mandated in all cases of pediatric strokes and TIAs unlike adults as more often than not there is the presence of some underlying vascular etiology of developmental origin. Early diagnosis and tailored management are needed in every case to reduce mortality and morbidity.

Declarations

Consent for publication: The author clarifies that written informed consent was obtained and the anonymity of the patient was ensured. This study submitted to Swiss J. Rad. Nucl. Med. has been conducted in accordance with the Declaration of Helsinki and according to requirements of all applicable local and international standards.
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Conflict of interest:

The authors declare that there were no conflicts of interest within the meaning of the recommendations of the International Committee of Medical Journal Editors when the article was written.

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