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Moyamoya Disease with Intraventricular Hemorrhage in a Child: A Case Report

Nisha Regmi[1], Bibek Shrestha[2], Vivek Karn[3], Pradeep Raj Regmi[4]

Author(s) affiliation

[1]*College of Medical Sciences, Chitwan, Nepal*

[2]*Maharajgunj Medical Campus, Tribhuvan University Teaching Hospital, Institute of Medicine, Kathmandu, Nepal*

[3]*Department of Pediatrics, Maharajgunj Medical Campus, Tribhuvan University Teaching Hospital, Institute of Medicine, Kathmandu, Nepal*

[4]*Department of Radiology, Maharajgunj Medical Campus, Tribhuvan University Teaching Hospital, Institute of Medicine, Kathmandu, Nepal*

Corresponding author

Bibek Shrestha, MBBS
shresthabibek85iom@gmail.com

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ABSTRACT

Moyamoya disease is a rare progressive cerebrovascular disorder in Nepal characterized by stenosis or occlusion of cerebral arteries with abnormal collateral vessels, and hemorrhagic presentation in children is extremely uncommon. We report an 8-year-old girl with intraventricular hemorrhage presenting with seizures, vomiting, and decreased consciousness. Imaging revealed characteristic angiographic features of Moyamoya disease. She was managed with external ventricular drainage and supportive treatment, achieving significant recovery. This case highlights the importance of considering Moyamoya disease in pediatric hemorrhagic stroke and underscores the need for early imaging and timely intervention.

Keywords
Moyamoya Disease; intraventricular hemorrhage; pediatric stroke

INTRODUCTION

Moyamoya disease is a progressive occlusive cerebrovascular disorder characterized by stenosis or occlusion of the internal carotid arteries and their major branches, leading to the development of fragile collateral vascular networks[1]. The disease is more commonly reported in East Asian populations but has been increasingly recognized worldwide. In pediatric patients, ischemic events such as transient ischemic attacks and strokes are the predominant presentations. However, hemorrhagic forms, though rare in children, pose significant diagnostic and therapeutic challenges[2]. We present an first reported case of intraventricular hemorrhage in an 8-year-old girl subsequently diagnosed with Moyamoya disease in Nepal.

CASE PRESENTATION

An 8-year-old female presented with a sudden onset occipital headache, vomiting, and transient loss of consciousness followed by increased limb tone, incontinence, and recurrent episodes of seizure-like activity. There was no history of trauma, fever, or prior similar events. On examination,

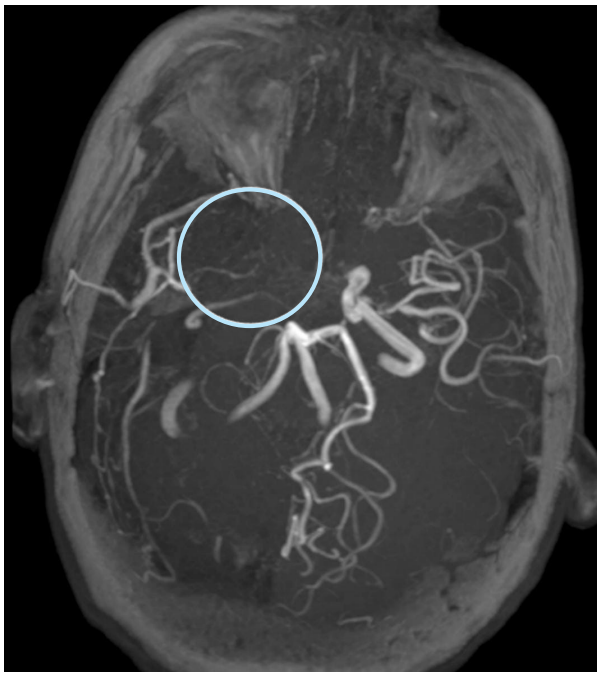

Figure 1. Magnetic resonance angiography of the circle of Willis demonstrating severe stenosis of the distal internal carotid artery with markedly attenuated proximal MCA and ACA branches. The circled region highlights a dense network of fine basal collateral vessels, producing the characteristic "puff-of-smoke" appearance typical of Moyamoya disease (blue circle).

she was unconscious (GCS 4/15), with sluggishly reactive pupils, decreased muscle tone, absent deep tendon reflexes, and unresponsive plantar reflexes. Vitals were stable, and there were no meningeal signs. Differential diagnoses included seizures, meningitis, encephalitis, metabolic derangements, ADEM, and vascular anomalies. Lab results were within normal limits. Magnetic Resonance Imaging (MRI) with angiography revealed T2 hyperintensities in the right periventricular and left centrum semiovale regions, with non-visualization of the right MCA and proximal A1 segment of ACA, along with paucity of cortical vessels—findings suggestive of Moyamoya disease. (Figure 1) Computed Tomography (CT) of the brain showed intraventricular hemorrhage with raised intracranial pressure. (Figure 2)

The patient underwent emergency external ventricular drainage via left Kocher's point; CSF was turbid and under high pressure. The turbid appearance of the CSF was attributed to the presence of blood degradation products due to intraventricular hemorrhage rather than infection, as microbiological analysis showed no growth and cytology revealed xanthochromic features consistent with hemorrhagic origin. Postoperatively, she managed with Vancomycin, Meropenem, Levetiracetam, and supportive care. Vancomycin was administered empirically as part of the prophylactic regimen to prevent ventriculostomy-related infection, which is a recognized complication following external ventricular drain placement in pediatric neurosurgical cases. Repeat imaging showed resolution of hemorrhage. On follow-up, the child had significant clinical improvement with no neurological deficits or seizures, confirming the diagnosis and effective management of Moyamoya disease presenting with intraventricular hemorrhage.

DISCUSSION

Moyamoya disease is a rare and progressive cerebrovascular disorder marked by the narrowing or occlusion of internal carotid arteries and their primary branches, leading to the formation of abnormal collateral vessels. [1,2] It primarily affects East Asian populations, with a higher prevalence in Korea and Japan compared to Western countries. The disease exhibits two incidence peaks: around age 10 and 30-45 years, with later onset in women. Children typically present with ischemic

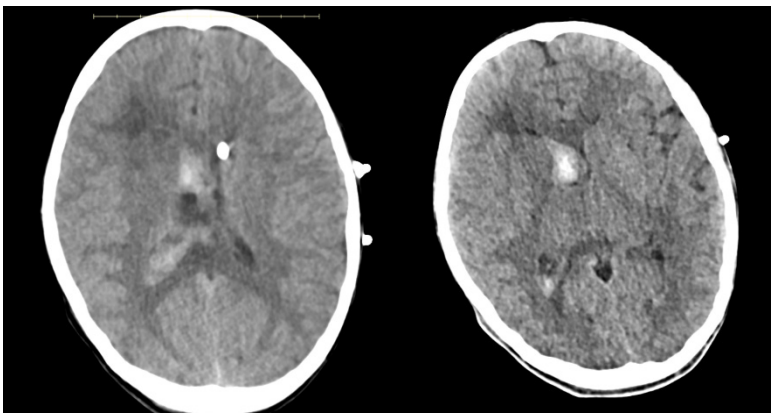

Figure 2. CT Brain: showing resolved intraventricular hemorrhage with external ventricular drain placement

symptoms, while adults more often experience intracranial hemorrhage. Genetic studies have identified RNF213 as a susceptibility gene for moyamoya disease, with the p.R4810K variant common in East Asian patients.[3] This variant was found in 56% of Asian-descent Moyamoya patients in a diverse U.S. population, while other RNF213 variants were identified in non-Asian patients.[4] Moyamoya is a rare cerebrovascular disorder with an incidence of 0.35-0.94 per 100,000 population, it primarily affects Asian populations but is also reported in other parts of the world.[5] Common symptoms include transient ischemic attacks, strokes, headaches, seizures, and sensorimotor paralysis. Herein the patient presented with the complaints of unconsciousness, and the seizure attacks. In children, moyamoya disease is often associated with other congenital conditions, such as neurofibromatosis type 1 and Down syndrome, which may complicate the diagnostic process.[2] The presence of intraventricular hemorrhage in a pediatric patient raises concerns about potential underlying vascular malformations or secondary moyamoya syndrome, which can occur in conjunction with other systemic conditions.[6] Radiological diagnosis plays a crucial role in screening, evaluating vascular changes, and clinical follow-up. MRI and MR angiography are considered the most reliable non-invasive methods for visualizing primary findings, such as arterial occlusion and collateral formation, as well as secondary findings like cerebral infarction and hemorrhage. CT with contrast enhancement can reveal tortuous vessels in the basal ganglia, corresponding to collaterals seen on angiography. CT angiography is also valuable in diagnosing vascular changes.[7] Here, MR angiogram was done which showed high intensity area in right periventricular white area and left centrum semiovale and non-visualization of right middle cerebral artery and proximal A1 segment of right anterior cerebral artery with paucity of cortical vessels on the ipsilateral side and confirmed the diagnosis of Moyamoya disease as a primary diagnosis. Moyamoya disease can lead to refractory high intracranial pressure, particularly following intraventricular hemorrhage, requiring aggressive management such as ventricular drainage, barbiturates, and hypothermia. High intracranial pressure is a critical concern, often requiring aggressive interventions such as ventricular drainage, barbiturates, and hypothermia.[8] While traditionally, experts advised against evacuating intraventricular hemorrhage in the acute phase, recent cases suggest that early surgical removal of severe IVH casting may be beneficial in managing critical ICP and improving outcomes.[9] In this case, decrease the elevated intracranial pressure and hydrocephalus, the patient underwent external ventricular drain placement. Along with raised ICP, seizures are a known complication, with 9-19% of pediatric moyamoya patients developing epilepsy.

CONCLUSION

This case emphasizes the need to include Moyamoya disease in the differential diagnosis of pediatric intraventricular hemorrhage. Early imaging and surgical intervention play key roles in management and recovery. Future protocols should aim at early recognition and tailored treatment strategies for hemorrhagic variants.

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CONSENT

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CONFLICT OF INTEREST

The author(s) declare that they do not have any conflicts of interest with respect to the research, authorship, and/or publication of this article.

AUTHOR CONTRIBUTIONS

BS: Conceptualization, Original writing-review and editing; VK and PR: Supervision

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