ECRONICON OPEN ACCESS

EC CLINICAL AND MEDICAL CASE REPORTS

Case Report

Two Challenges, One Child: Sickle Cell Disease and Moyamoya in a Pediatric Patient

H Qajia*, C Jabbari, S El Kadiri, N Eccherif El Kettani, F Touarsa, M Jiddane and M Fikri

Department of Neuroradiology, Mohamed V University, Rabat, Morocco

***Corresponding Author:** H Qajia, Department of Neuroradiology, Mohamed V University, Rabat, Morocco.

Received: May 05, 2025; **Published:** June 19, 2025

Abstract

Moyamoya disease is a rare cerebrovascular condition marked by progressive narrowing of the internal carotid artery and the circle of Willis, leading to the formation of collateral vessels in the basal ganglia. While the exact cause remains unknown, Moyamoya syndrome is associated with conditions like neurofibromatosis type I, down syndrome, radiation exposure, and, less commonly, sickle cell disease. Sickle cell disease, a genetic disorder causing abnormal hemoglobin production and red blood cell deformation, can lead to stroke and vascular complications. This report discusses a rare case of a 5-year-old boy with sickle cell disease who developed left-sided weakness and aphasia at age 2. Brain MRI revealed sequelae of anoxic-ischemic encephalopathy and characteristic "puff of smoke" collateral vessels indicative of Moyamoya. The patient was treated conservatively and discharged with minimal improvement in weakness, highlighting the rare association of Moyamoya with sickle cell disease.

Keywords: *Sickle Cell Disease; Moyamoya Syndrome; Pediatric*

Introduction

Sickle cell disease (SCD) results from a mutation in the beta-globin chain of hemoglobin (HbS), leading to abnormal polymerization under physiological stress such as hypoxia, hyperosmolarity, infection, and acidosis. This promotes erythrocyte sickling, contributing to vaso-occlusion, hemolytic anemia, ischemia-reperfusion injury, hypercoagulability, and systemic inflammation [1]. The resulting vascular damage increases the risk of cerebrovascular accidents (CVA), with an age-adjusted incidence of 0.61 per 100 person-years in homozygous SCD (HbSS) [2]. Cerebrovascular complications in SCD include ischemic and hemorrhagic strokes, silent infarcts, dural venous thrombosis, and Moyamoya syndrome (MMS) [3]. Moyamoya vasculopathy is a progressive condition involving intracranial arterial stenosis, prompting collateral vessel formation. Reduced cerebral blood flow in the internal carotid arteries (ICAs) and their branches predisposes patients to recurrent strokes and transient ischemic attacks [4]. MMS is diagnosed based on characteristic vascular patterns seen on magnetic resonance angiography (MRA), often associated with SCD and other conditions such as neurofibromatosis type I, medulloblastoma, down syndrome, and renal artery stenosis [5]. We present a case of MMS in a child with SCD, highlighting key considerations for stroke diagnosis and management in SCD patients.

Case Report

A 5-year-old boy, born to first-degree consanguineous parents and diagnosed with sickle cell disease (SCD), presented with intellectual disability, developmental delay, and behavioral disturbances. His medical history was significant for an ischemic stroke at the age of 2, resulting in left-sided hemiplegia and aphasia.

On examination, he exhibited persistent left hemiparesis with reduced spontaneous movement and muscle strength in both the left upper and lower limbs. Grip strength was weaker on the left side, while deep tendon reflexes were normal. Cognitive assessment revealed significant intellectual impairment.

Laboratory tests, including blood and cerebrospinal fluid analyses, were unremarkable. Electroencephalography (EEG) showed poor background activity. Cardiac evaluation, including echocardiography and electrocardiography, revealed no structural or functional abnormalities.

A brain MRI was performed to assess the sequelae of anoxic-ischemic encephalopathy. Imaging revealed a right porencephalic cyst with peripheral gliosis and atrophy of the corpus callosum, consistent with anoxic-ischemic injury (Figure 1). Additionally, there was severe stenosis of the supra-clinoid portion of both internal carotid arteries, with absent flow in the anterior and middle cerebral arteries, along with the development of Moyamoya collaterals in a "puff of smoke" pattern (Figure 2). These findings were consistent with Moyamoya syndrome, contributing to chronic ischemic changes.

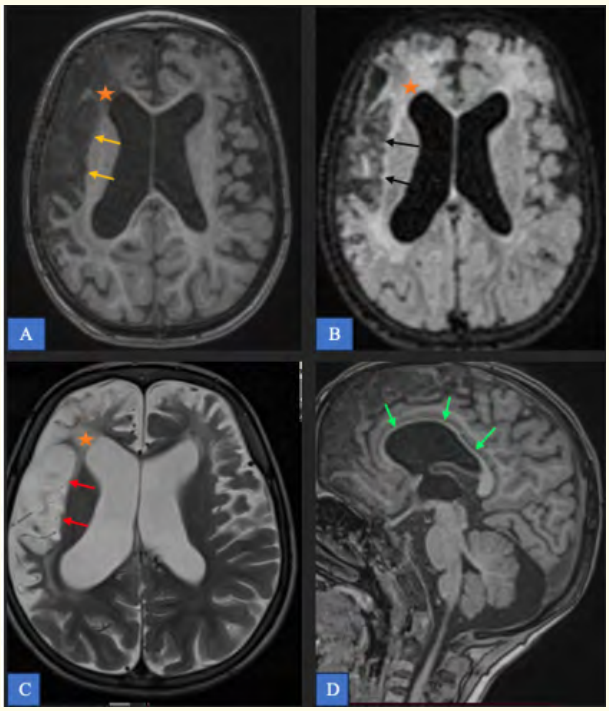

Figure 1: *A-C: Axial T1 (A), FLAIR (B), and T2-weighted imaging (C) showing a right fronto-parieto-temporal porencephalic cyst, appearing hypointense on T1 (yellow arrows), hypointense on FLAIR (black arrows), and hyperintense on T2 (red arrows), with associated peripheral gliosis (orange asterisk) suggestive of anoxic sequelae. D: Sagittal T1 MRI slice demonstrating atrophy of the corpus callosum (green arrows) in the context of an anoxic-ischemic injury.*

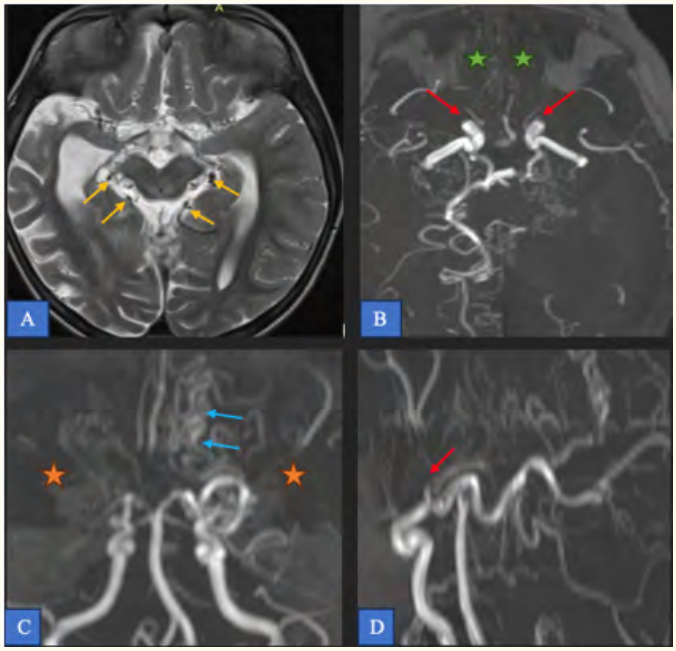

Figure 2: *A: T2 axial weighted imaging of MRI showing multiple flow voids related to collateral vessels called moyamoya vessels (yellow arrows). B-D: Axial (B), coronal (C), and sagittal (D) TOF (time of flight) MRA showing severe stenosis of the supra-clinoid portion of both internal carotid arteries (red arrows), with absent flow in the anterior (green asterisks) and middle cerebral arteries (orange asterisks), and the development of a moyamoya collaterals in a "puff of smoke" pattern (blue arrows).*

The patient was managed conservatively with supportive care and neurological follow-up. Despite intervention, there was minimal improvement in left-sided weakness.

Discussion

Sickle cell disease (SCD) is a known risk factor for Moyamoya disease. The chronic hemolysis and vaso-occlusion in SCD lead to vascular stress, which promotes the development of collateral blood vessels in the brain, characteristic of MMD. These abnormal vessels are seen on imaging as the “puff of smoke” appearance and help compensate for impaired cerebral blood flow. The combination of these vascular abnormalities increases the risk of ischemic strokes, particularly in children [6].

Moyamoya disease (MMD) is a rare cerebrovascular disorder characterized by progressive narrowing of the terminal portions of the internal carotid arteries and the circle of Willis, leading to reduced blood flow to the brain and ischemic events [7]. The cause of MMD remains unclear, while moyamoya syndrome (MMS) refers to a similar vascular pathology resulting from conditions such as autoimmune disorders, neurofibromatosis type I, down syndrome, radiation exposure, and rarely, and rarely sickle cell disease [8]. Diagnostic imaging for MMD relies on angiographic abnormalities, such as stenosis of the internal carotid arteries (ICA) and/or proximal vessels of the circle of Willis, alongside the development of prominent basal or parenchymal collaterals (Moyamoya vessels), visible in angiography, magnetic resonance angiography (MRA), or computed tomography angiography (CTA) [7].

Cerebral angiography is the gold standard for diagnosing MMD/MMS [2]. This invasive procedure provides detailed information on the carotid and cerebral arteries, including collateral circulation. Characteristic findings include occlusion or stenosis of the supraclinoid ICA segment and extensive collateral vessels in the parenchyma, transdural, and leptomeningeal regions. In conventional angiography, these abnormal vessels appear as a "puff of smoke" [9]. MMD progression can be classified in stages: (i) narrowing of the ICA bifurcation, (ii) initiation of basal Moyamoya vessels, (iii) increased Moyamoya vessels with the "puff of smoke" appearance, (iv) further increase of collateral vessels, (v) narrowing of ICAs and destruction of Moyamoya vessels, and (vi) complete occlusion of ICAs with collateral development from the external carotid artery (ECA) [10].

Recent MRI advancements have greatly enhanced the ability to diagnose MMD. MRI can reveal ICA stenosis or occlusion, Moyamoya vessels, brain ischemia, infarction, atrophy, and ventriculomegaly. Various MR sequences, including FLAIR, DWI, perfusion, and contrast-enhanced imaging, are used for diagnosis [11]. T2W images are optimal for visualizing stenotic arteries, while FLAIR images highlight parenchymal changes. FLAIR and post-contrast T1W images can show leptomeningeal involvement, indicated by the "ivy sign," which reflects decreased cerebrovascular reserve. DWI detects small infarcts, and gradient echo sequences reveal chronic microbleeds [11]. MRA, using the 3D-TOF technique, offers non-invasive, accurate mapping of the ICA and ECA without the need for gadolinium contrast, making it ideal for post-operative assessment [12].

Surgical intervention is essential for symptomatic patients, particularly in children. The primary goal of surgery is to prevent ischemic and hemorrhagic strokes by bypassing affected arteries or creating new blood flow sources [13]. Early diagnosis and revascularization generally lead to a more favorable prognosis.

Conclusion

Moyamoya disease is a rare cerebrovascular disorder that can occur as a primary condition or as part of MMS. In children with SCD, clinicians must consider non-embolic causes of stroke, such as MMS. Early diagnosis is essential to improve outcomes and prevent further neurological deterioration.

Bibliography

1. Shah F and Dwivedi M. "Pathophysiology and recent therapeutic insights of sickle cell disease". *Annals of Hematology* 99.5 (2020): 925-935.
2. Ohene-Frempong K., *et al*. "Cerebrovascular accidents in sickle cell disease: rates and risk factors". *Blood* 91.1 (1998): 288-294.
3. Webb J and Kwiatkowski JL. "Stroke in patients with sickle cell disease". *Expert Review of Hematology* 63 (2013): 301-316.
4. Scott RM and Smith ER. "Moyamoya disease and moyamoya syndrome". *New England Journal of Medicine* 360.12 (2009): 1226-1237.
5. Soares D., *et al*. "Moyamoya syndrome in sickle cell anaemia: a cause of recurrent stroke". *BMJ Case Reports* (2014): bcr2014203727.
6. Koh JW., *et al*. "Moyamoya disease in sickle cell disease: A review". *Journal of Stroke and Cerebrovascular Diseases* 26.8 (2017): 1790-1796.
7. Mimi L., *et al*. "Moya Moya disease: about 3 cases and a review of literature". *Diagnostic and Interventional Imaging* 2.1 (2019): 22-31.
8. Tavares Bello C., *et al*. "Down syndrome and Moyamoya disease: an unusual cause of stroke". *BMJ Case Reports* (2017): bcr-2017-219894.
9. Odriozola EA., *et al*. "Radiological findings in Moya Moya" (2013).

10. Hertza J., *et al*. "Moyamoya disease: a review of the literature". *Applied Neuropsychology: Adult* 21.1 (2014): 21-27.

11. Osborn AG., *et al*. "Diagnostic imaging: brain. 3rd edition". Philadelphia, PA 19103-2899: Elsevier. ISBN: 978-0-323-37754-6 chapter 81: Moyamoya (2016): 294-297.

12. Yamada I., *et al*. "Moyamoya disease: comparison of assessment with MR angiography and MR imaging versus conventional angiography". *Radiology* 196.1 (1995): 211-218.

13. Kim T., *et al*. "Moyamoya disease: treatment and outcomes". *Journal of Stroke* 18.1 (2016): 21-30.

Volume 8 Issue 7 July 2025