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CASE REPORT

Posterior ischemic optic neuropathy as a complication of Moyamoya syndrome in a 36-year-old male with sickle cell disease

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Abstract

Purpose: To describe a unique case of posterior ischemic optic neuropathy (PION) in a 36-year-old man with sickle cell disease (SCD) and Moyamoya syndrome (MMS), highlighting the rare and complex interaction of these conditions and their implications in ocular ischemic events.

Case Report: A North African male with homozygous SCD presented with sudden, painless diminution of vision in his left eye, 2 months before consulting in our department. On examination, his visual acuity was preserved in the right eye but limited to counting fingers at one meter in the left eye. A significant left relative afferent pupillary defect was noted. Funduscopic examination and Fluorescein angiography showed signs of proliferative sickle cell retinopathy, with optic disc atrophy in the left eye. Computed tomography angiography and magnetic resonance angiography of the brain confirmed MMS with stenosis of the internal carotid artery and Moyamoya vessel formation, as well as optic nerve atrophy. No recovery in visual acuity was observed one year later.

Conclusion: This case is notable for the unusual coexistence of SCD, MMS, and PION in the same patient. It also highlights the complexity of managing coexisting SCD and MMS, as both conditions amplify the risk of cerebral and ocular ischemic events, leading to irreversible vision loss. Multidisciplinary care is essential to monitor and address the diverse manifestations of these overlapping conditions and prevent severe outcomes.

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Introduction

Sickle cell disease (SCD) is a hematological disorder characterized by abnormal hemoglobin, which predisposes individuals to a variety of vascular complications, including ocular and neurological ischemic events, due to sickling of red blood cells [1]. Moyamoya syndrome (MMS) is a rare vasculopathy characterized by progressive stenosis of the internal carotid arteries (ICAs) and the development of fragile collateral vessels, which increases the risk of cerebral ischemic events in SCD patients [2].

Although ischemic optic neuropathy is a rare complication in SCD, it can occur in the form of anterior ischemic optic neuropathy (AION) or posterior ischemic optic neuropathy (PION). PION results in sudden diminution of vision due to ischemia of the retrobulbar optic nerve, and is typically associated with systemic hypotension or severe vascular occlusions. It is particularly rare in SCD, where ischemic events are more commonly seen in the retinal vasculature [3,4].

The coexistence of SCD, MMS, and PION in a single patient presents a unique and complex clinical challenge, amplifying the risk of both cerebral and ocular ischemic events. This case report explores the unusual combination of these three conditions in a 36-year-old man, highlighting the intricate interaction between hematological, vascular, and

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neurological factors that contribute to the patient's severe clinical presentation.

Case Report

A 36-year-old North African man with a history of SCD (SS genotype) presented to the emergency department for a sudden, painless diminution of vision in his left eye. He reported numerous previous episodes of transient monocular vision loss in the same eye, with three distinct episodes occurring in the month preceding the permanent vision loss. The first two episodes lasted only a few minutes and resolved spontaneously. The last episode, occurring 2 weeks before the persistent vision loss, lasted for 4 days and resolved gradually, but with a slow and incomplete recovery. However, as the current episode persisted without any improvement, the patient sought medical attention 2 months after the onset of symptoms.

On examination, his best-corrected visual acuity was 20/20 in the right eye but was limited to counting fingers at one meter in the left eye. A pronounced relative afferent pupillary defect was noted in the left eye. External eye examination and applanation tonometry were unremarkable, with normal conjunctival vessels and a clear anterior segment observed on slit-lamp examination. Funduscopic examination revealed bilateral signs of sickle cell retinopathy, including salmon patch hemorrhages and veinous tortuosity. In the left eye, optic disc atrophy was observed, while the right optic disc appeared normal (Fig. 1).

Fluorescein angiography demonstrated signs of nonproliferative sickle cell retinopathy in the right eye (Fig. 2) and proliferative sickle cell retinopathy in the left eye. The left eye exhibited extensive peripheral retinal non-perfusion, diffuse narrowing of retinal arterioles, and characteristic arteriovenous anastomoses at the ischemic borders (Fig. 3). Computed tomography angiography revealed narrowing of the high cervical and intracavernous segments of the left ICA (Fig. 4). Magnetic resonance angiography of the brain revealed stenosis and occlusion of the terminal portion of the left ICA, affecting both its cervical and intracranial segments (Fig. 5). Collateral vessels, known as Moyamoya vessels, were detected along the left ophthalmic artery, along with diffuse atrophy of the left optic nerve (Fig. 6). Narrowing of the left middle cerebral artery with rarefaction of the insular cortical circulation was also objectified in the 3D TOF sequence (Fig. 7). Evidence of a previous ischemic junctional stroke on the left side was also found on axial T2 and FLAIR sequences (Fig. 8). There was no improvement in the visual acuity on follow up examination 1 year later and no further progression of the retinopathy.

Discussion

The present case is unusual due to the rare and complex association of three distinct conditions: SCD, MMS, and PION. While each of these conditions can individually cause vascular ischemic events, their coexistence in a single patient amplifies the risk and complexity of ischemic damage.

SCD is caused by a mutation in the beta globin chain of hemoglobin, leading to the formation of abnormal hemoglobin. Under physiological stress, such as hypoxia, infection, or acidosis, hemoglobin S polymerizes, causing red blood cells to take on a sickled shape. These sickled cells lead to vaso-occlusion, hemolytic anemia, ischemia-reperfusion injury, hyper-coagulability, and systemic inflammation [1]. The degree of vascular injury and



Figure 1. Fundus photographs revealing bilateral signs of sickle cell retinopathy: veinous tortuosity (arrowhead in a) and salmon patch hemorrhages (arrow in b). Presence of an optic disc atrophy in the left eye (arrowhead in b).

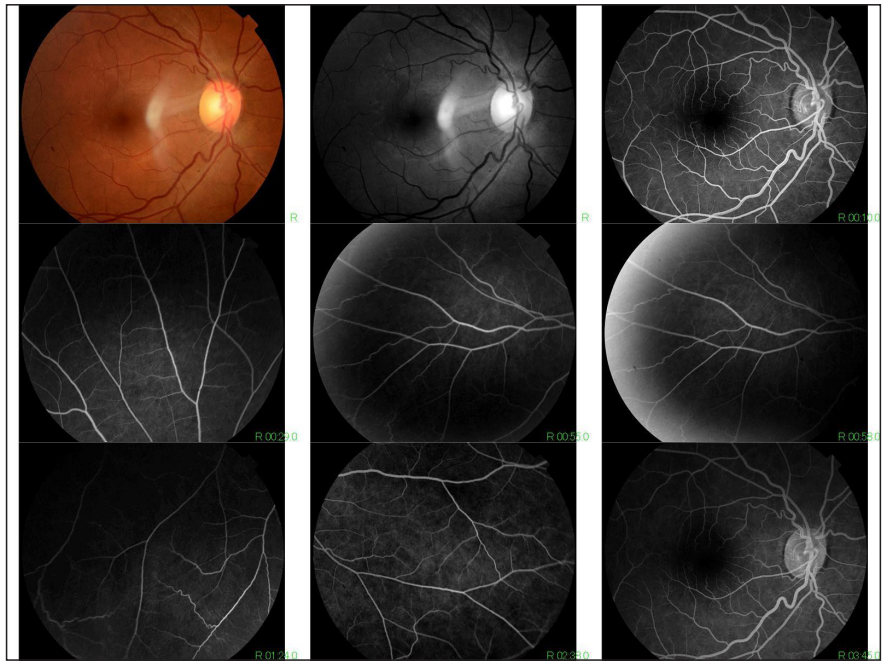


Figure 2. Fluorescein angiography of the right eye demonstrating signs of nonproliferative sickle cell retinopathy.



Figure 3. Fluoresceine angiography of the left eye showing signs of proliferative sickle cell retinopathy stage 2 of Goldberg: diffuse arteriolar narrowing, ischemic retinal territories in the far periphery (arrowhead) and arteriovenous anastomoses (arrow).

inflammation in SCD predisposes patients to a higher rate of cerebral vascular accidents [5]. The mechanisms behind these strokes are varied and include ischemic and hemorrhagic events, silent infarcts, dural venous thrombosis, and in some cases, MMS [6,7].

In this patient, the ischemia is further intensified by MMS, a progressive occlusive vasculopathy that leads to stenosis of the terminal portion of the ICAs

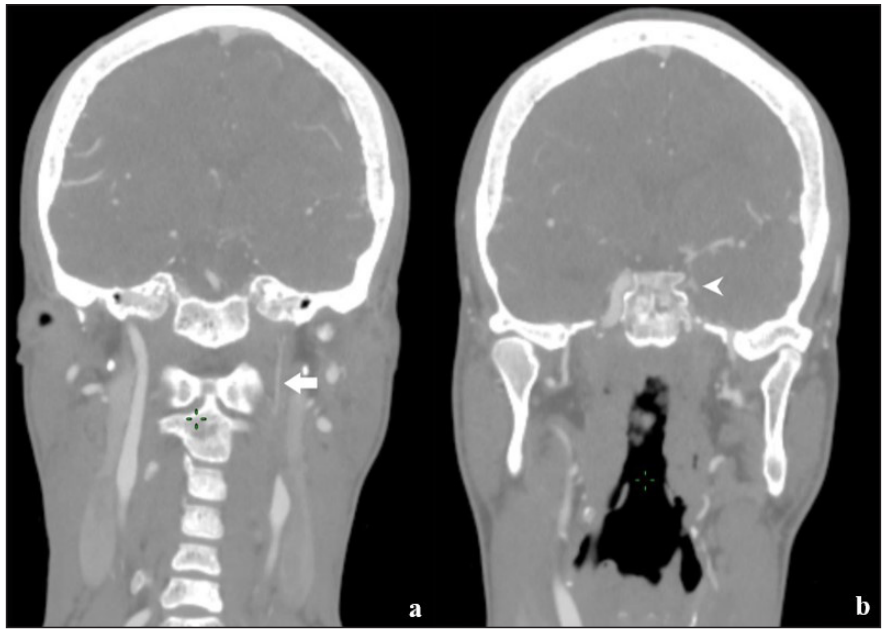


Figure 4. Coronal MIP CT scans revealing a narrowed appearance of the high cervical portion of the left ICA (arrow in a), with a lack of opacification of its intracavernous segment (arrowhead in b).

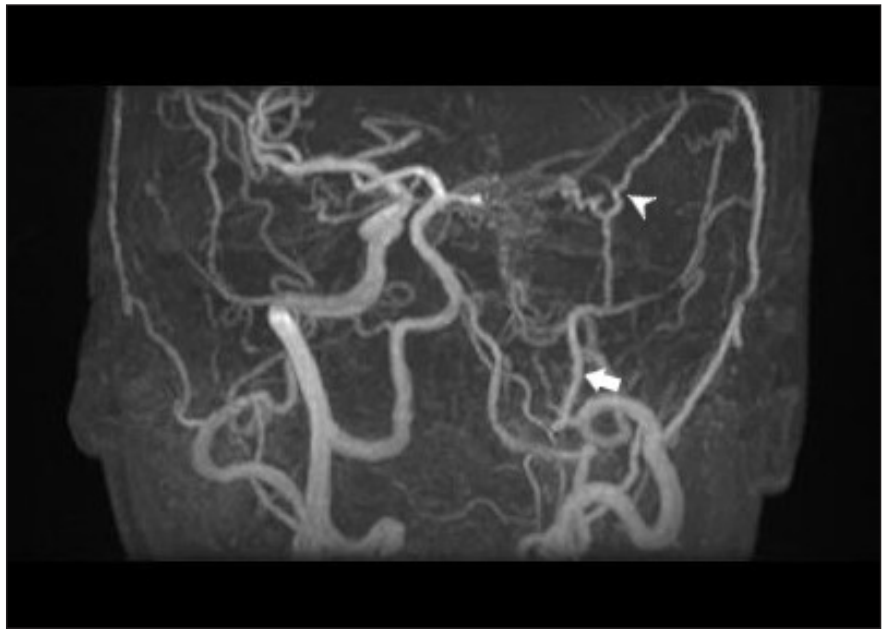


Figure 5. Reconstruction of angiographic sequences revealing a narrowed appearance of the left ICA (arrow), with a lack of opacification of its high cervical, intrapetrous, and intracavernous segments, with a thin revascularization of its supraclinoid portion. This also shows a narrowed appearance of the middle cerebral artery (arrowhead).

and the development of fragile collateral vessels, called Moyamoya vessels [2]. Epidemiologically, this condition occurs mostly in children and rarely in adults [8]. Moyamoya disease refers to the disease itself without a predisposing condition. However, MMS refers to Moyamoya in the presence of a predisposing condition, such as neurofibromatosis, systemic lupus erythematosus, tuberous sclerosis, and SCD [9,10]. MMS is a relative uncommon



Figure 6. 3D TOF sequence showing the absence of opacification of the intracavernous segment of the left ICA (arrow) and collateral circulation in the path of the left ophthalmic artery (arrowhead).

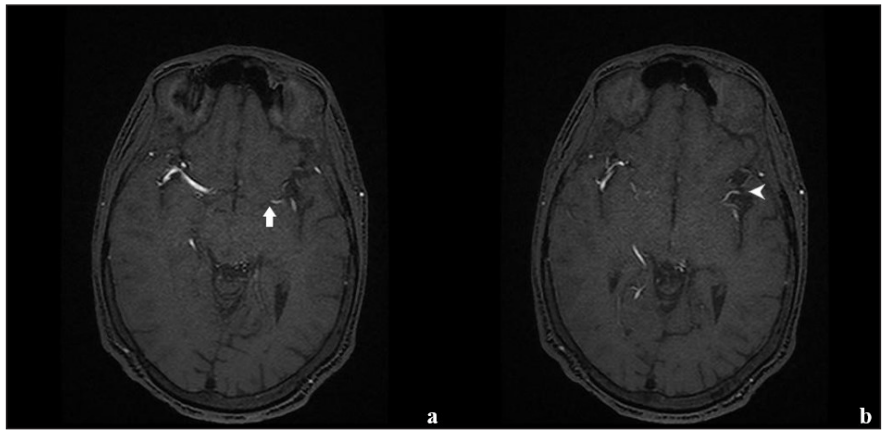


Figure 7. 3D TOF sequence showing a narrow appearance of the left middle cerebral artery (arrow in a) with rarefaction of the insular cortical circulation (arrowhead in b).

complication of SCD but significantly increases the risk of both cerebral and ocular ischemic events [11].

Acute vaso-occlusive events are common systemic manifestations of SCD, and ocular involvement is no exception. While retinal and choroidal vaso-occlusion is well-documented, optic nerve involvement remains rare [3,4]. The diagnosis of ischemic optic neuropathy in this case presents a challenge, particularly in distinguishing between AION and PION. While AION typically manifests with optic disc edema at the onset of vision loss, this clinical feature could not be assessed in our patient, as he sought medical attention 2 months after the onset of symptoms. By that time, optic disc atrophy was already present, making it impossible to confirm or rule out initial disc swelling. However, several factors support a diagnosis of PION rather than AION. The evidence of severe cervical and intracranial ICA stenosis, strongly suggests impaired

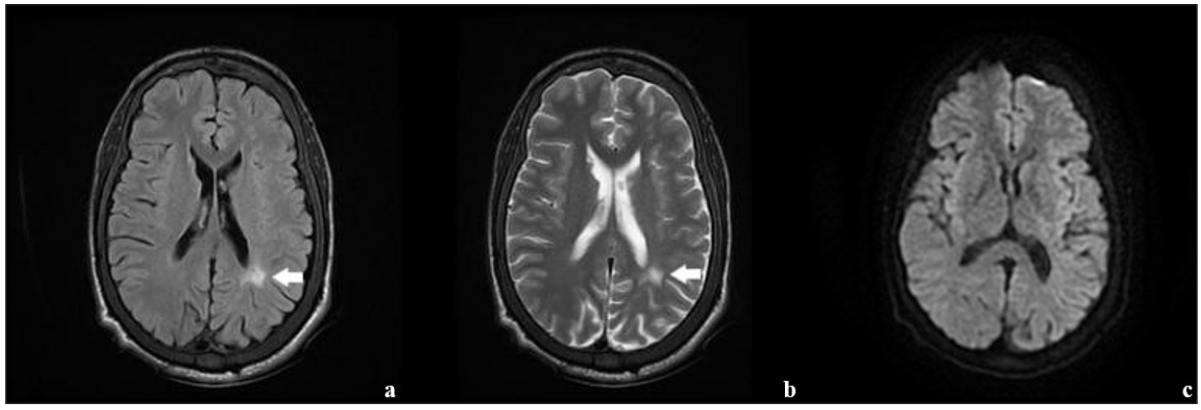


Figure 8. Axial MRI sequences showing a lesion adjacent to the occipital horn of the left lateral ventricle, appearing hyperintense on T2 and FLAIR (arrow in a and b), with no corresponding abnormality on diffusion sequence (c), consistent with a chronic ischemic stroke.

posterior ciliary artery circulation, which supplies the retrobulbar portion of the optic nerve. In contrast, AION is usually linked to small vessel disease or giant cell arteritis rather than large artery occlusions like those seen in this case. Furthermore, the presence of collateral Moyamoya vessels and sickle cell vasculopathy point to a more proximal vascular compromise, affecting blood supply beyond the optic disc, which aligns with the pathophysiological mechanisms underlying PION. Therefore, despite the lack of early fundoscopic findings, the overall clinical and radiological findings strongly favor PION as the cause of vision loss in this patient.

PION is a rare optic neuropathy where ischemia affects the retrobulbar portion of the optic nerve, usually linked to systemic hypotension, perioperative events, or vascular occlusions. It typically presents with profound visual loss and a relative afferent pupillary defect without acute disc swelling. The distinct arterial supply of the posterior optic nerve by independent collateral of the ophthalmic artery, which is independent of the anterior segment, explains why the optic disc may appear normal in the acute phase, with optic atrophy developing later [4]. Although AION has been reported in association with ocular migraines, this patient did not report any pain or headache with the onset of visual loss, further supporting a PION diagnosis [3,12].

The patient's recurrent episodes of transient monocular vision loss further support the diagnosis of PION. These episodes, particularly the longer ones lasting hours to days, suggest deeper ischemia with slow recovery, indicating a progressive optic neuropathy and an important risk of irreversible damage. In the context of MMS, hypoperfusion of the posterior optic nerve, makes it susceptible to transient ischemic episodes, while the fragile collateral Moyamoya vessels provide unstable and insufficient blood flow, exacerbating fluctuations in perfusion and further compromising optic nerve integrity [9].

Additionally, the patient exhibited signs of sickle cell retinopathy, a known complication in SCD, characterized by salmon patch hemorrhages, venous tortuosity, and neovascular formations resembling sea fans. However, the central visual loss in this patient is best explained by the posterior ischemic event rather than retinal ischemia alone.

The management of MMS in SCD patients can include medical or surgical management. Endovascular surgery, particularly encephaloduroarteriosynangiosis, has been shown to reduce the risk of recurrent strokes, especially in pediatric populations [13]. Medical management often involves chronic blood transfusions to reduce sickling, with or without hydroxyurea [14].

The simultaneous presence of these three conditions makes this case highly unusual, as it highlights the challenging interaction between hematological, vascular, and neurological factors, leading to profound and irreversible vision loss. This stresses the need for a multidisciplinary approach to managing such complex interactions.

Conclusion

This case highlights the complex interaction between SCD and MMS in the development of PION. The patient's history of SCD predisposed him to recurrent ischemic episodes, while the additional MMS exacerbated the vascular insufficiency leading to optic nerve damage. PION remains a rare but devastating complication in SCD patients, especially

when coupled with systemic vascular anomalies such as MMS.

Early recognition and close monitoring of ocular and cerebral vascular complications in SCD are essential to prevent irreversible outcomes like vision loss. This case emphasizes the need for multidisciplinary management to address both ocular and systemic manifestations of these coexisting conditions.

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None.

Conflicts of interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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Ethics approval

This study was conducted in accordance with the principles of the Declaration of Helsinki.

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