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Case Report

Subtle signs, serious consequences: pediatric moyamoya disease presenting with seizures and behavioural regression

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ABSTRACT

Moyamoya disease (MMD) is a rare, progressive cerebrovascular arteriopathy characterized by chronic stenosis or occlusion of the terminal internal carotid arteries with compensatory development of a fragile basal collateral network. Although classically described with well-recognized presentations such as stroke, transient ischemic attacks, or seizures, its clinical spectrum in young children is remarkably varied, and early manifestations are often subtle, nonspecific, and easily overlooked or misattributed to more common childhood conditions. This diagnostic ambiguity is compounded in resource-limited settings, where ready access to vascular neuroimaging is not always available, and where a low index of suspicion for a rare cerebrovascular disorder can result in children being managed symptomatically for weeks or months before the underlying diagnosis is considered. We report a 2-year-2-month-old female who presented with a three-month history of behavioural change and regression of previously attained expressive language, followed by transient right-sided weakness, recurrent right-sided focal motor seizures, and progressive hemiparesis. Her early symptoms of irritability and language regression were initially unrecognized as neurological in origin, and even her first episode of transient limb weakness resolved without further evaluation, illustrating how easily the earliest, subtlest features of this disease can be missed. It was only with the onset of recurrent seizures and persistent motor deficits that neuroimaging was pursued. Magnetic resonance imaging demonstrated an acute infarct in the left inferior frontal and parietal regions, while magnetic resonance angiography revealed marked bilateral narrowing of the terminal internal carotid, anterior, and middle cerebral arteries with extensive basal collaterals producing the classical "puff-of-smoke" appearance. Non-contrast computed tomography additionally revealed chronic left hemispheric encephalomalacia with gyriform calcification, indicating an earlier, clinically silent ischemic insult that had gone unrecognized. Digital subtraction angiography confirmed bilateral terminal internal carotid artery stenosis, graded predominantly as Suzuki stage III. The child underwent left-sided encephaloduroarteriosynangiosis (EDAS) with good postoperative recovery of hemiparesis. This case emphasizes that Moyamoya disease must be considered even in the presence of subtle, seemingly unrelated symptoms such as behavioural change or language regression, particularly in settings where diagnostic delay due to varied age-specific presentation and limited imaging access can allow silent, cumulative ischemic injury before diagnosis, underscoring the need for heightened clinical vigilance across all pediatric age groups.

Keywords: Moyamoya disease, Pediatric stroke, Cerebral arteriopathy, Encephaloduroarteriosynangiosis, Magnetic resonance angiography, Diagnostic delay, Suzuki Staging

INTRODUCTION

Moyamoya disease (MMD) is a chronic, progressive, non-atherosclerotic cerebrovascular disorder characterized by idiopathic stenosis or occlusion of the terminal portions of the internal carotid arteries and their proximal branches, resulting in the formation of a fine network of fragile basal collateral vessels. On cerebral

angiography, these collateral vessels produce the characteristic hazy appearance resembling a "puff of smoke," from which the disease derives its Japanese name, moyamoya.[1,2]

MMD occurs either as an isolated entity (Moyamoya disease) or in association with underlying conditions such as neurofibromatosis type 1, sickle cell disease, Down syndrome, and cranial irradiation, in which case it is referred to as Moyamoya syndrome. The disease shows a bimodal age distribution, with one peak in childhood (around 5-10 years) and a second in the fourth decade of life, though onset in infancy and toddlerhood is well documented and tends to follow a more aggressive course.[2,3] While it remains most prevalent in East Asia-with reported incidence rates of 0.94 per 100,000 in Japan and 2.3 per 100,000 in South Korea, compared with under 0.1 per 100,000 in Western populations-increasing numbers of cases are now being reported from South Asia and other historically low-incidence regions, suggesting the true burden in these areas may be underestimated due to underdiagnosis rather than true rarity.[3-5]

Children with Moyamoya disease typically present with ischemic manifestations, including transient ischemic attacks (TIAs), arterial ischemic stroke, and recurrent episodes of hemiparesis, which may be precipitated by hyperventilation during crying, fever, or exertion. Seizures are increasingly recognized as both a presenting and accompanying manifestation, particularly in children with established cerebral infarction.[6,7] In addition, chronic cerebral hypoperfusion may result in cognitive impairment, behavioural disturbances, and language regression, especially in patients with early-onset disease.[7,8] Critically, the clinical phenotype of MMD varies considerably across age groups: infants and toddlers more often present with subtle, nonspecific features such as irritability, developmental regression, or isolated behavioural change, whereas older children more typically present with overt, easily recognizable focal deficits or stroke-a distinction that can cause the disease to go unrecognized for prolonged periods when it presents at the younger end of the spectrum.[7-9]

The diagnosis of MMD is frequently delayed because its presenting features often mimic more common pediatric neurological disorders such as primary epilepsy, postictal paresis, cerebral palsy, or neurodevelopmental disorders, and subtle early symptoms-such as irritability or language regression-are easily overlooked or attributed to behavioural causes rather than triggering neurological evaluation. This diagnostic challenge is further compounded in resource-limited settings, where access to advanced vascular neuroimaging such as MRA or DSA may be delayed or unavailable at the primary point of contact, and where the varied, age-dependent presentation of the disease demands a correspondingly high index of clinical suspicion to avoid missed or delayed diagnosis.[5,10] Consequently, children may experience multiple, sometimes clinically silent, ischemic events before vascular imaging establishes the diagnosis. Early recognition is crucial, as timely initiation of antiplatelet therapy and surgical revascularization can substantially reduce the risk of recurrent stroke and improve long-term neurological outcomes.[7,11]

We report a toddler who initially presented with subtle behavioural change and language regression, followed by transient ischemic episodes and recurrent focal seizures, before being diagnosed with Moyamoya disease. This case emphasizes that Moyamoya disease should be considered even when presenting symptoms are subtle and nonspecific, and highlights the diagnostic challenges posed by its varied, age-dependent presentation, particularly in resource-limited settings where delayed recognition can allow silent, cumulative ischemic injury before diagnosis.

CASE REPORT

A 2-year-2-month-old female child, the firstborn of a non-consanguineous couple, presented with a three-month history of behavioural change, recurrent seizures, transient episodes of right-sided weakness, and progressive right hemiparesis. She was born at term by lower-segment caesarean section with a birth weight of 2.7 kg. Her antenatal, natal, and neonatal periods were uneventful, with no history of birth asphyxia, neonatal seizures, jaundice, or neonatal intensive care unit admission. Developmental milestones were appropriate for age until the onset of her illness.

Three months before presentation, the parents noticed a gradual change in the child's behaviour, characterized by excessive crying, irritability, and aggressive outbursts. These symptoms were initially attributed to temper tantrums. Over the following weeks, she developed progressive regression of expressive language. Previously, she had been able to speak in two- to three-word sentences appropriate for her age, but gradually lost this ability and became unable to speak meaningful words. There was no history of fever, vomiting, head trauma, altered sensorium, or other systemic illness during this period.

Approximately one month after the onset of behavioural symptoms, she developed a sudden episode of weakness involving the right upper limb, associated with decreased spontaneous movements. The weakness resolved completely within 4-5 hours following symptomatic treatment at a local hospital. In retrospect, this episode was suggestive of a transient ischemic attack.

Fifteen days later, she experienced another episode of sudden-onset weakness involving both the right upper and lower limbs. Around the same time, the parents observed recurrent episodes of focal motor seizures involving the right upper limb, accompanied by head deviation, upward rolling of the eyes, and frothing from

the mouth. Each episode lasted approximately 20-30 seconds and was not associated with urinary or fecal incontinence, tongue bite, or prolonged postictal confusion. She also experienced occasional generalized tonic-clonic seizures over the following weeks, prompting further neurological evaluation and hospital admission. Magnetic resonance imaging (MRI) of the brain demonstrated diffusion restriction with corresponding T2/FLAIR hyperintensity involving the left inferior frontal and parietal regions, consistent with an acute ischemic infarction (Figure 1). No abnormal post-contrast enhancement was noted. The remaining cerebral parenchyma, ventricular system, posterior fossa structures, and major intracranial flow voids appeared unremarkable.

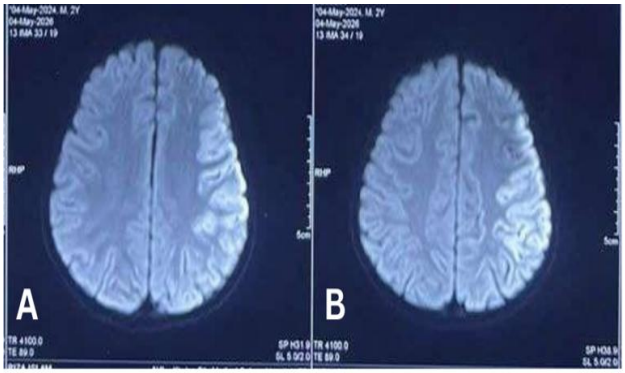

Figure 1 (A and B): Axial MRI brain (diffusion/FLAIR sequence) showing restricted diffusion with signal change in the left inferior frontal and parietal regions, in keeping with an acute infarct.

Time-of-flight magnetic resonance angiography (MRA) demonstrated marked narrowing of the terminal segments of both internal carotid arteries, extending to the circle of Willis and the proximal anterior and middle cerebral arteries bilaterally. Moderate narrowing of both posterior cerebral arteries was also noted. Extensive collateral vessels were visualized within the bilateral capsuloganglionic regions, producing the characteristic "puff-of-smoke" appearance, with no evidence of aneurysmal dilatation or other vascular malformations (Figure 2). These findings were diagnostic of bilateral Moyamoya disease.

Non-contrast computed tomography (CT) of the brain demonstrated diffuse hypodensity involving the left frontoparietotemporal cortex, subcortical white matter, and deep periventricular white matter, along with widening of the cortical sulci, prominence of the left Sylvian fissure, gyriform dystrophic calcification, and mild left cerebral hemiatrophy (Figure 3). These findings were consistent with chronic encephalomalacic changes secondary to previous ischemic injury, indicating that the child had sustained earlier ischemic insults before the acute presentation. Following these imaging findings, the child was referred to our tertiary care centre for further management. At presentation, she was receiving levetiracetam syrup (1 ml twice daily) and aspirin 75 mg on alternate days. She was hemodynamically stable, with a pulse rate of 105 beats/minute, blood pressure of 100/62 mmHg, oxygen saturation of 97% on room air, weight of 9 kg, and height of 84 cm.

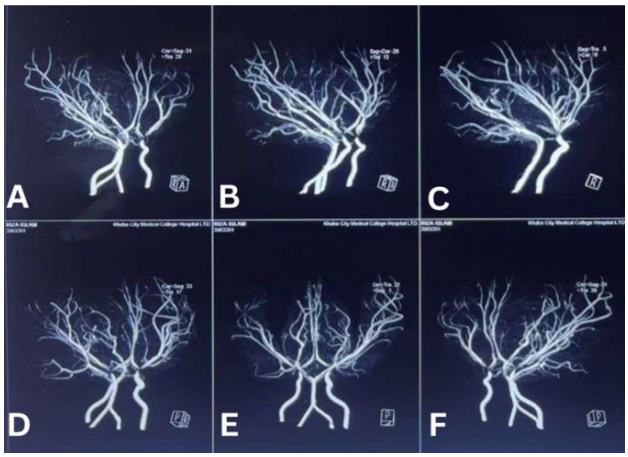

Figure 2: (A-F) 3D time-of-flight MR angiogram showing bilateral terminal ICA, ACA and MCA narrowing with an extensive fine collateral vascular network producing the classical "puff-of-smoke" appearance.

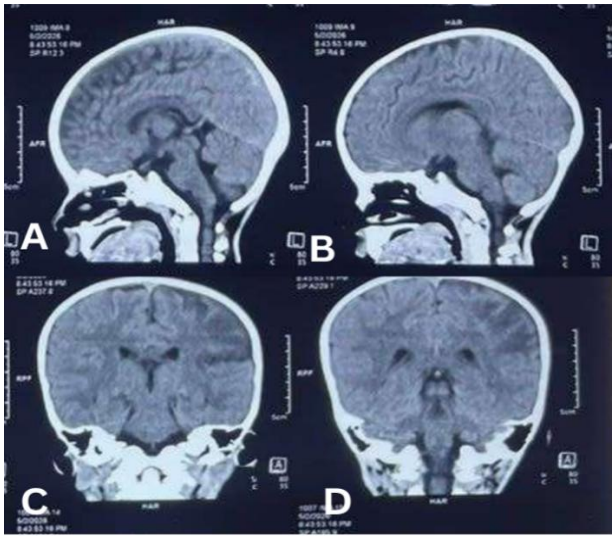

Figure 3: (A-D) Non-contrast CT brain (coronal and sagittal section) showing encephalomalacic change with gyriform (dystrophic) calcification and mild hemiatrophy of the left fronto-parieto-temporal region, reflecting chronic ischaemic injury.

Neurological examination revealed persistent irritability with excessive crying. Mild spasticity was present in the right upper and lower limbs. She was able to lift the right upper limb against gravity, although spontaneous movements were reduced. Deep tendon reflexes were preserved bilaterally, with an extensor plantar response on the right and a flexor plantar response on the left, consistent with an upper motor neuron lesion involving

the left cerebral hemisphere. The remainder of the systemic examination was unremarkable.

Digital subtraction angiography (DSA) confirmed bilateral stenosis of the terminal internal carotid arteries with extensive basal collateral vessel formation, graded predominantly as Suzuki stage III, consistent with the diagnosis of bilateral Moyamoya disease (Figure 4).

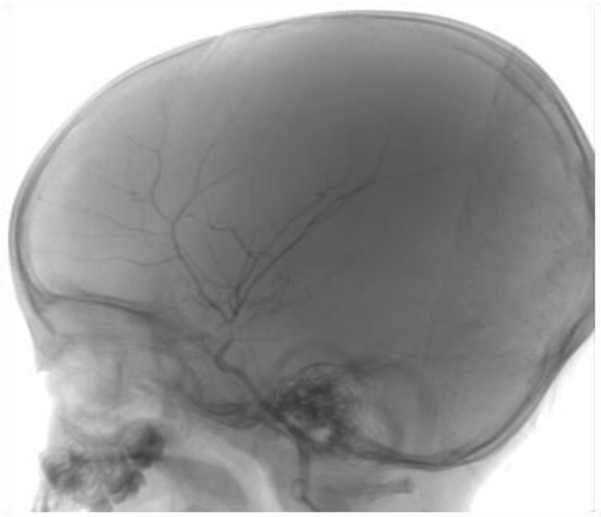

Figure 4: Digital subtraction angiogram of the left internal carotid artery, lateral view, showing sparse, attenuated filling of the distal anterior and middle cerebral artery branches with an extensive fine network of abnormal basal collateral vessels producing the characteristic "puff-of-smoke" appearance, findings in keeping with Suzuki stage III Moyamoya disease.

In view of symptomatic bilateral Moyamoya disease with established left hemispheric infarction and progressive right hemiparesis, the child underwent left-sided encephaloduroarteriosynangiosis (EDAS). The perioperative and postoperative periods were uneventful, with stable neurological and hemodynamic parameters throughout the hospital stay. She was continued on antiplatelet therapy and antiepileptic medication and was discharged in stable condition.

At follow-up, the child demonstrated gradual improvement in right-sided motor function, with noticeable recovery in limb power compared with her preoperative status. No further focal neurological deterioration or recurrent seizures were observed. She remains on regular neurological and neurosurgical follow-up, with staged revascularization of the contralateral hemisphere planned based on future clinical and radiological assessment.

DISCUSSION

Moyamoya disease is a rare, progressive cerebrovascular disorder characterized by chronic stenosis or occlusion of the terminal internal carotid arteries and their proximal branches, with the formation of fragile collateral vessels that compensate for progressive cerebral hypoperfusion. In children, ischemic events predominate, whereas adults more commonly present with intracranial hemorrhage. Early diagnosis is crucial, as timely surgical revascularization can significantly reduce the risk of recurrent ischemic injury and improve long-term neurological outcomes.[1,2]

This case highlights the diagnostic challenges associated with pediatric Moyamoya disease. The child initially presented with behavioural change and progressive language regression, followed by transient episodes of right-sided weakness and recurrent focal motor seizures. Because these manifestations are individually far more common in primary epilepsy, postictal paresis, or neurodevelopmental disorders, the diagnosis of an underlying cerebrovascular disease may easily be overlooked. Recognition of the transient neurological deficits and progression of symptoms prompted vascular neuroimaging, which established the diagnosis before further irreversible ischemic injury occurred.

Seizures are increasingly recognized as an important presenting feature of pediatric Moyamoya disease. Gatti et al. reported seizures in approximately 40% of children with Moyamoya arteriopathy and identified cerebral infarction as an important risk factor for their occurrence.[6] Similarly, Grigore et al observed that ischemic stroke and transient ischemic attacks remain the most frequent presenting manifestations in children, while seizures may occur either at presentation or later during the disease course, particularly in those with established ischemic injury.[7] In our patient, recurrent focal motor seizures were most likely secondary to cortical ischemia involving the left frontal and parietal regions rather than a primary epileptic disorder.

Behavioural disturbances and developmental regression are less commonly recognized manifestations of pediatric Moyamoya disease but may occur as a consequence of chronic cerebral hypoperfusion and repeated ischemic insults. Progressive language regression in our patient preceded the diagnosis and corresponded with imaging evidence of both acute infarction and chronic left hemispheric encephalomalacia. Similar presentations have been described by Habes et al who reported a toddler presenting with recurrent seizures and developmental regression prior to the diagnosis of Moyamoya disease.[8] These findings emphasize that unexplained developmental regression, particularly when accompanied by focal neurological deficits or seizures, should prompt evaluation for an underlying structural or vascular etiology.

Neuroimaging plays a pivotal role in establishing the diagnosis. MRI demonstrated an acute infarction involving the left inferior frontal and parietal regions,

while CT revealed chronic encephalomalacic changes with gyriform calcification and mild cerebral hemiatrophy, indicating previous ischemic injury. Time-of-flight MR angiography demonstrated bilateral terminal internal carotid artery stenosis with extensive basal collateral vessels producing the characteristic "puff-of-smoke" appearance. Digital subtraction angiography further confirmed these findings and graded the disease as Suzuki stage III, an intermediate angiographic stage characterized by intensification of the basal collateral network alongside clear narrowing of the major cerebral arteries. The coexistence of chronic encephalomalacia and acute infarction suggests that the child had sustained multiple ischemic events before the diagnosis was established, underscoring the progressive nature of the disease.

The revised 2021 diagnostic criteria permit a diagnosis of definite Moyamoya disease in patients with characteristic bilateral MRI and MRA findings without necessarily requiring catheter angiography.[10] Nevertheless, digital subtraction angiography remains the reference standard for defining vascular anatomy and staging disease severity, and is particularly valuable when surgical revascularization is being planned. In our patient, DSA confirmed bilateral terminal internal carotid artery stenosis (Suzuki stage III) with extensive collateral vessel formation, supporting definitive diagnosis and operative planning.

The cornerstone of management in pediatric Moyamoya disease is cerebral revascularization, which aims to improve cerebral perfusion and prevent recurrent ischemic events. Medical therapy with antiplatelet agents may reduce the risk of further cerebral ischemia but does not halt disease progression. Indirect revascularization procedures such as encephaloduroarteriosynangiosis (EDAS) have demonstrated favorable outcomes in children because of their greater angiogenic potential compared with adults.[7,11] Our patient underwent left-sided EDAS, with gradual postoperative improvement in right-sided motor function and no further neurological deterioration during follow-up, highlighting the benefits of timely surgical intervention even at an intermediate angiographic stage.

This case also reinforces an important clinical message. In toddlers presenting with recurrent focal seizures, transient or recurrent focal neurological deficits, behavioural change, or unexplained language regression, clinicians should maintain a high index of suspicion for cerebrovascular disorders, particularly when symptoms evolve over time or fail to fit a typical epilepsy syndrome. Early MRI combined with vascular imaging can facilitate prompt diagnosis, timely referral for neurosurgical intervention, and prevention of irreversible neurological disability.

CONCLUSION

Moyamoya disease, although rare, should be considered in toddlers presenting with recurrent focal seizures, transient or recurrent focal neurological deficits, and unexplained behavioural or language regression. Because these manifestations frequently mimic primary seizure disorders or neurodevelopmental conditions, diagnosis may be delayed until irreversible cerebral ischemic injury has occurred. Early recognition with MRI and vascular imaging enables timely initiation of antiplatelet therapy and referral for surgical revascularization, thereby reducing the risk of recurrent stroke and improving long-term neurological outcomes. This case emphasizes the importance of maintaining a high index of suspicion for cerebrovascular disease in young children with evolving focal neurological symptoms.

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