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Case of Moya Moya Disease

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Abstract: ***Moyamoya disease is a chronic, progressive cerebrovascular disorder characterized by stenosis of the internal carotid arteries and development of compensatory collateral vessels, leading to ischemic or hemorrhagic events. We report a case of a 03 - year - old female presenting with weakness of left upper and lower limb, ultimately diagnosed with Moyamoya disease. Neuroimaging revealed bilateral arterial narrowing and collateral vessel formation. This case underscores the importance of early recognition of neurological symptoms in young children and timely surgical intervention to prevent long - term morbidity.***

Keywords: “Moyamoya disease", "pediatric stroke", Magnetic resonance imaging, Computed tomography

1. Introduction

Moyamoya disease is a rare, progressive cerebrovascular disorder characterized by idiopathic stenosis or occlusion of the supraclinoid internal carotid arteries and their major branches, particularly the anterior and middle cerebral arteries. This leads to the formation of fragile collateral vessels at the base of the brain, which appear as a "puff of smoke" (Moyamoya in Japanese) on angiography, giving the disease its name. First described in Japan in the 1960s, Moyamoya disease is now recognized worldwide, with a higher prevalence in East Asian populations (incidence of 3–10 per 100, 000 in Japan versus 0.5–1 per 100, 000 in Western countries). The condition exhibits a bimodal age distribution, with peaks in childhood (5–10 years) and early adulthood (30–40 years), though cases in children under 5 years are uncommon and pose unique diagnostic challenges.

The pathophysiology of Moyamoya disease remains incompletely understood, but genetic and environmental factors are implicated. Mutations in the *RNF213* gene have been strongly associated with the disease, particularly in East Asian cohorts, suggesting a hereditary predisposition. In pediatric patients, the disease typically presents with ischemic symptoms, such as transient ischemic attacks (TIAs), seizures, or strokes, due to chronic hypoperfusion. In contrast, adults are more likely to experience intracranial hemorrhage from rupture of fragile collaterals. The progressive nature of the disease can lead to significant morbidity, including cognitive impairment and motor deficits, if untreated.

Diagnosis relies heavily on neuroimaging. Magnetic resonance angiography (MRA) and digital subtraction angiography (DSA) are critical for visualizing arterial stenosis and collateral networks, with DSA remaining the gold standard for staging disease severity using the Suzuki classification. Early recognition is essential, as delayed diagnosis increases the risk of irreversible neurological damage. Treatment strategies focus on restoring cerebral blood flow, with surgical revascularization—either direct or indirect—being the mainstay for symptomatic patients. In children, indirect procedures like encephaloduroarteriosynangiosis (EDAS) are often preferred due to technical challenges with small vessels.

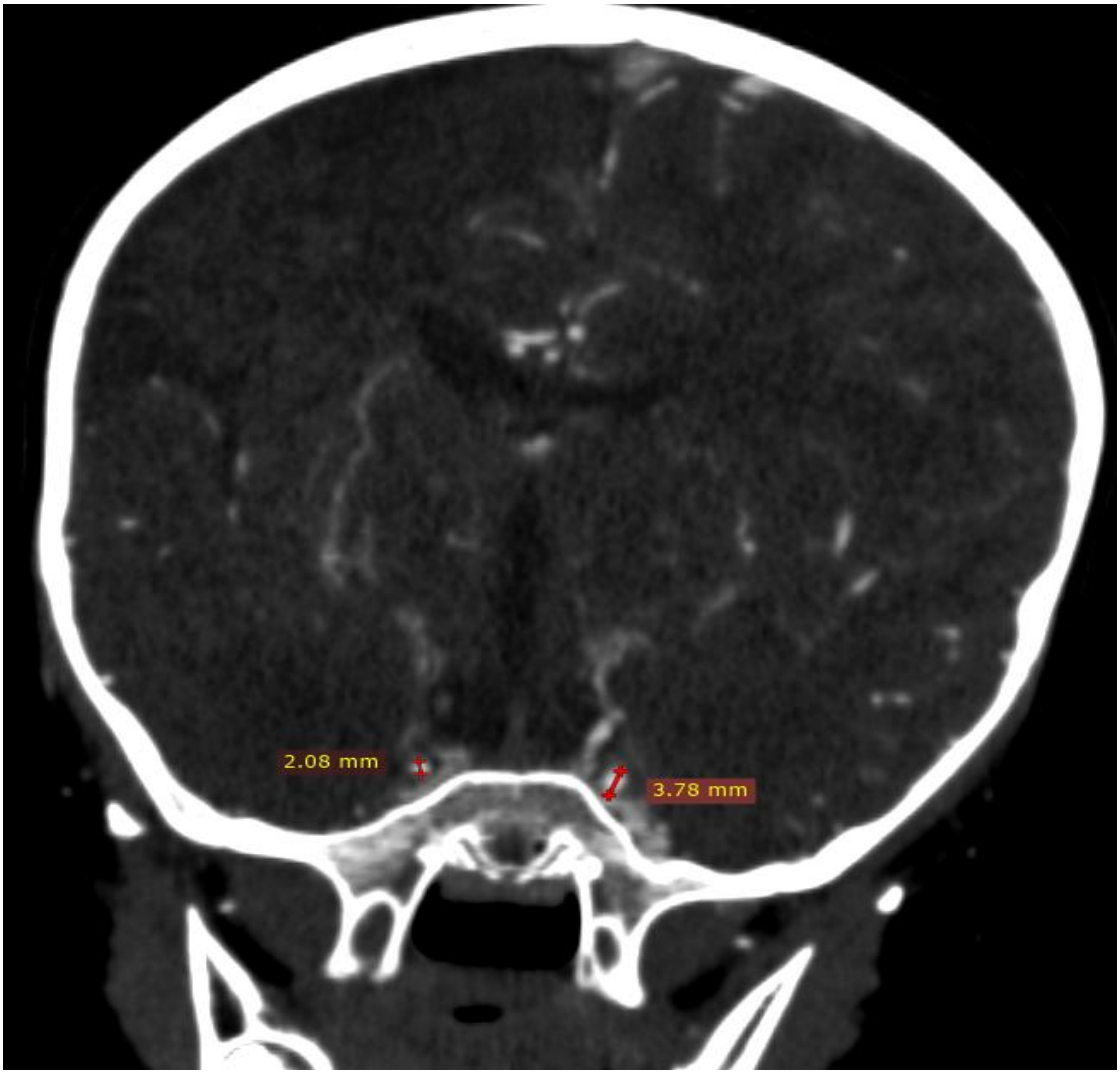
Pediatric Moyamoya disease presents unique challenges, including atypical symptoms that may mimic developmental disorders or epilepsy, leading to diagnostic delays. Recent studies emphasize the importance of heightened clinical suspicion in young children with recurrent neurological symptoms, as early intervention can significantly improve long - term outcomes. This case report describes the clinical course of a 3 - year - old girl diagnosed with Moyamoya disease after presenting with TIAs, highlighting the critical role of timely diagnosis and surgical management in preventing further ischemic events.

2. Case Presentation

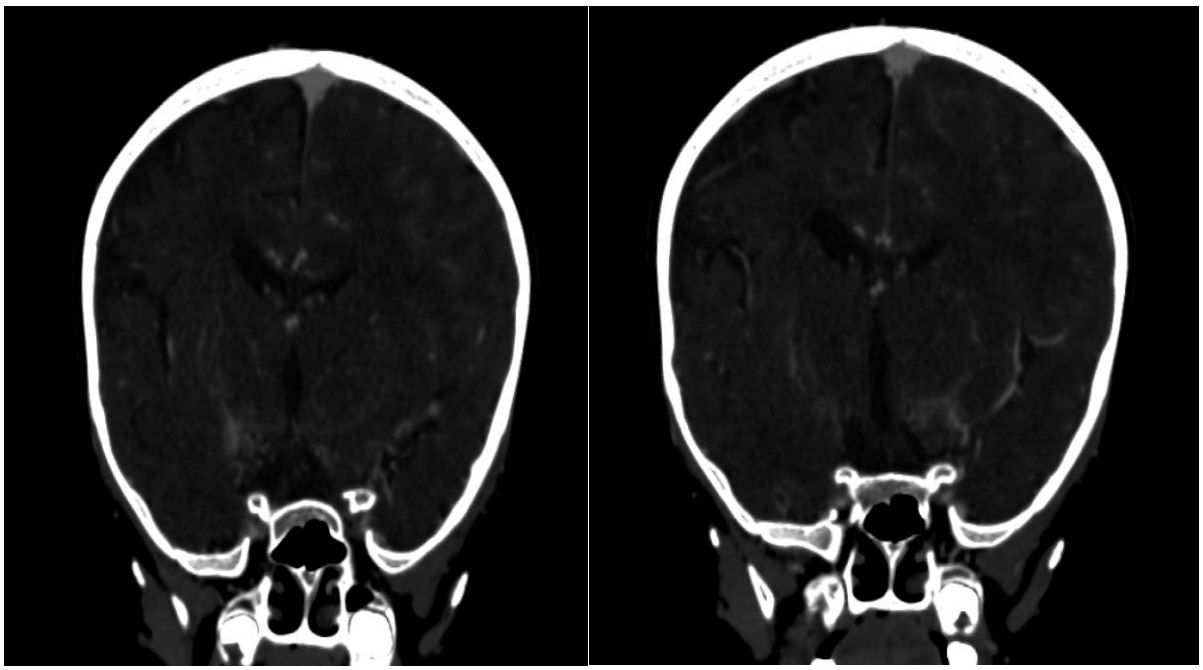
A 3 - year - old previously healthy girl presented to the pediatric emergency department with a 2 - month history of recurrent episodes of left sided weakness and difficulty speaking, each lasting 10–20 minutes with full recovery between episodes. Her parents reported three such episodes, with the most recent accompanied by a fall. There was no history of seizures, fever, head trauma, or developmental delay. The family history was unremarkable, with no known neurological or vascular disorders.

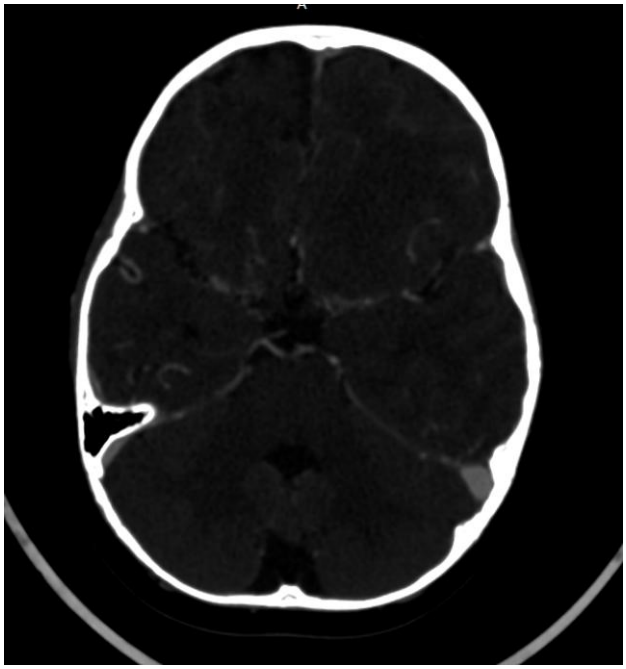
On examination, the patient was alert and afebrile, with normal vital signs (blood pressure 95/60 mmHg, heart rate 100 bpm, respiratory rate 20/min). Neurological examination during an asymptomatic period was normal, with no focal deficits, normal tone, and age - appropriate developmental milestones. No neurocutaneous markers, facial asymmetry, or cardiac murmurs were noted. Initial laboratory tests, including complete blood count, electrolytes, and coagulation profile, were within normal limits. Cerebrospinal fluid (CSF) analysis was not performed due to the absence of infectious or inflammatory signs.

3. Imaging Findings



Bilateral (right > left) internal carotid arteries appear narrow throughout their entire course with decreasing caliber from its origin to end. With maximum 80 - 90 %luminal compromise predominantly in the supraclinoid segments





Tuft of tortuous collaterals also noted in the right gangliocapsular region, right temporo - parietal lobes and bilateral (right > left) parafalcine parietal lobes.

4. Discussion

Although more prevalent in East Asia, Moyamoya disease should be considered in young adults presenting with unexplained TIAs or strokes (3). MRI and angiographic imaging are essential for diagnosis (4). Management includes medical therapy for stroke prevention and surgical revascularization for symptomatic patients, which has shown favorable outcomes in reducing ischemic events (5, 6).

5. Conclusion

This case underscores the need for high clinical suspicion for Moyamoya disease in young adults with recurrent neurological symptoms. Early diagnosis and surgical management can significantly improve prognosis and quality of life.

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