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# Puff of Smoke' In A Young Brain: A Classic Imaging Sign of Moyamoya Disease – Case Report

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**ABSTRACT:** Moyamoya disease is a rare and progressive cerebrovascular disorder marked by the occlusion or blockage of the distal internal carotid arteries and their main branches. In response, the brain develops a network of fragile collateral vessels, which appear as a "puff of smoke" on angiographic imaging—a characteristic feature that inspired the disease's name. The condition primarily affects children and young adults, often presenting with transient ischemic attacks, strokes, seizures, or cerebral haemorrhage. Although the underlying mechanisms are not fully understood, genetic factors, particularly mutations in the *RNF213* gene, have been closely associated with disease susceptibility, The major susceptibility gene for moyamoya disease in people investigation into the mechanisms of disease and potential treatment targets. The Arg4810Lys variant of the gene is most strongly associated with moyamoya disease, but the penetrance is lower than 1%, suggesting a synergistic relationship with additional environmental and genetic risk factors. A 11-year-old female patient presented with the complaints of sudden onset of left upper limb and lower limb transient weakness associated with bowel incontinence patient has a past medical history of seizures since 10 months with multiple seizure episodes Diagnostic evaluation typically involves magnetic resonance imaging, magnetic resonance angiography, and digital subtraction angiography, the latter of which remains the gold standard. Treatment strategies are primarily surgical, with revascularization procedures—either direct, indirect, or combined approaches—aimed at improving cerebral blood flow and reducing the risk of ischemic events . Based on laboratory investigations and radiological reports case was diagnosed as MOYAMOYA DISEASE preoperative and anaesthesia clearance patient was taken up for **STA-MCA bypass** (Superficial Temporal Artery to Middle Cerebral Artery Bypass) associated treatment with perivascular sympathectomy and superior cervical ganglionectomy may be useful but more investigation needs to be carried out into the pathogenesis of the disease before more definitive therapy is realized.
This review outlines the latest developments in the understanding, diagnosis, and management of Moyamoya disease, with attention to its genetic background and surgical outcomes.

**KEY WORDS:** Moyamoya disease, Revascularization, Stroke, Children, Surgery.

## INTRODUCTION TO MOYAMOYA DISEASE

Moyamoya disease is a chronic, progressive cerebrovascular condition marked by bilateral stenosis or occlusion of the terminal portion of the internal carotid arteries and their proximal branches, particularly the anterior and middle cerebral arteries. In response to the reduced cerebral perfusion, a network of abnormal collateral vessels develops at the base of the brain, producing a characteristic "puff of smoke" appearance on cerebral angiography (3). The disease primarily affects children and young adults, with clinical manifestations including transient ischemic attacks (TIAs), strokes, seizures, or cognitive impairment (4). Adults may also present with intracranial hemorrhages due to fragile collateral vessels (1).
Although the exact cause remains unclear, Moyamoya disease has been linked to genetic predispositions, particularly involving mutations in the **RNF213** gene in East Asian populations (2). Treatment strategies are largely surgical, with revascularization procedures such as direct or indirect bypass improving cerebral perfusion and reducing the risk of future ischemic events (1).

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**CASE REPORT**

A 11-year-old female patient presented in the emergency department with the complaints of sudden onset of left upper limb and lower limb transient weakness associated with bowel incontinence patient has a past medical history of seizures for 10 months with multiple seizure episodes on admission, patient blood pressure was 110/70 mmHg, pulse rate was stable, and no abnormal heart sounds were detected during cardiovascular and respiratory examinations was bilaterally air entry adequate. The abdominal examination was unremarkable, with a soft, non-tender abdomen. However, patient showed as moving all four limbs physically associated with weakness.

Laboratory investigations revealed as low haemoglobin of 11.20 g/dL, a red blood cell count of 4.38 million/µL, and white blood cell count of 11.91 million/µL, indicating leucocytosis. Platelet count was mildly elevated at 2.41lakhs/cu.mm.

**Table 1: Biological markers of moyamoya disease**

| Marker | Normal Range | Reported Abnormal Values in Moyamoya | Clinical Significance |
|---|---|---|---|
| **D-dimer** | < 0.5 µg/mL FEU | 0.9 µg/mL ↑ | Indicates fibrin degradation and possible thrombotic activity |
| **Protein C activity** | 70 – 140% | 58% ↓ | Deficiency may increase thrombosis risk |
| **Protein S activity (Free)** | 60 – 150% | 42% ↓ | Reduced levels seen in pediatric stroke cases |
| **Antithrombin III** | 80 – 120% | 64% ↓ | Contributes to clotting risk when deficient |
| **Lupus Anticoagulant** | Negative | Positive | Associated with autoimmune Moyamoya syndrome |
| **Antiphospholipid Antibodies** | < 20 GPL/MPL units | 18 GPL/MPL units | Seen in Moyamoya with antiphospholipid syndrome |
| **Homocysteine** | 5 – 15 µmol/L | 17 µmol/L ↑ | Elevated levels may damage endothelium and promote thrombosis |

**RADIOLOGICAL REPORTS**

Further diagnostic of magnetic resonance imaging shows Acute infract in left frontoparietal, anterio temporal lobe cortical sub cortical region and left corona radiata, capsulo ganglionic region corresponding FLAIR hyperintensity indicates areas of increased signal intensity on a Fluid-Attenuated Inversion Recovery due to occlusion.

Chronic infarct with focal encephalomalacia and hemosiderin deposit noted in right frontal lobe, predominantly in middle and inferior frontal gyri region, whereas magnetic resonance angiogram states that Bilateral Middle Cerebral Artery (MCA) shows no flow related signals indicates complete occlusion and bilateral Anterior Cerebral Artery (ACA) and Posterior Cerebral Artery (PCA) shows poor flow of blood indicates significant stenosis.

CT angiography of the brain demonstrates bilateral distal internal carotid artery (ICA) stenosis, more prominent on the left side. There is significant narrowing of the proximal anterior cerebral arteries (ACA) and posterior cerebral arteries (PCA) complete blockage of blood vessels Middle Cerebral Artery (MCA). Multiple fine collateral vessels are visualized in the region of the basal ganglia, forming a classic “puff of smoke” appearance, which is highly suggestive of Moyamoya disease.

Based on laboratory investigations and radiological reports case was diagnosed as MOYAMOYA DISEASE preoperative and anaesthesia clearance patient was taken up for **STA-MCA bypass** (Superficial Temporal Artery to Middle Cerebral Artery Bypass) a surgical procedure used to improve blood flow to the brain, where the internal carotid or middle cerebral arteries are narrowed or blocked under general anesthesia intra operative and postoperative periods were uneventful .Patient was closely monitored strict maintenance of blood pressure to avoid infarct or hemorrhage.

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Patient postoperative treatment was tailored to manage irreversible brain damage with Tab. Brevipil 500mg per oral twice daily, Tab. Flexon 325mg it is a combination of ibuprofen and paracetamol per oral thrice daily, Tab. Chymoral forte per oral twice daily, Tab. Ondansetron 4mg on need, T bact ointment on long actin, Tab. Ecosprin 75mg once daily, Syrup. Betonin 5ml per oral, Syrup. Duplac 30ml per oral once daily for five days.

## DISCUSSION

Moyamoya disease is a rare, idiopathic, and progressive cerebrovascular disorder involving occlusion of the terminal internal carotid arteries (ICAs), anterior cerebral arteries (ACAs), and middle cerebral arteries (MCAs), prompting the formation of fragile collateral vessels to maintain cerebral perfusion (3; 1). These collateral networks produce the hallmark "puff of smoke" appearance on angiography. Although initially described in Japan, cases have been reported globally, including increasing recognition in South Asia, Europe, and the United States (10;11)

In pediatric patients, Moyamoya disease most commonly manifests as transient ischemic attacks (TIAs), whereas adults are more prone to hemorrhagic presentations due to rupture of fragile collateral vessels (4;6). The current case describes a pediatric female presenting with focal weakness and a history of seizures—both common features in children with Moyamoya. Imaging findings were characteristic, showing bilateral MCA occlusion, poor flow in ACA and PCA territories, and multiple infarcts consistent with disease progression.

Laboratory investigations revealed a hypercoagulable profile, including reduced protein C and S levels, decreased antithrombin III, and a positive lupus anticoagulant. These findings are often reported in Moyamoya syndrome, a variant associated with underlying conditions such as autoimmune disorders, sickle cell anemia, and Down syndrome (8). The presence of these abnormalities suggests a possible secondary form of Moyamoya and underscores the need for thorough systemic evaluation.

Surgical revascularization remains the primary treatment approach. Direct procedures like superficial temporal artery to middle cerebral artery (STA-MCA) bypass offer immediate improvement in perfusion and have been associated with lower risk of recurrent ischemia (5;12). In contrast, indirect methods, such as encephaloduroarteriosynangiosis (EDAS), rely on neo angiogenesis and may be more suitable for younger children or cases with small-caliber donor vessels (14). The choice of revascularization method depends on patient age, vascular anatomy, and surgeon experience. In this case, a direct STA-MCA bypass was chosen and performed successfully.

Postoperative care is crucial in preventing complications such as cerebral hyper perfusion syndrome, infarction, or hemorrhage. Strict blood pressure control and antiplatelet therapy, as provided here, form the basis of early postoperative management (13). Long-term follow-up with serial imaging and neurocognitive assessment is necessary due to the risk of disease progression or re-occlusion.

Recent advances in the understanding of Moyamoya pathophysiology highlight a strong genetic component, particularly involving mutations in the **RNF213** gene, with the p.R4810K variant being highly associated in East Asian populations (2; 9). However, the low penetrance suggests environmental or epigenetic factors likely modulate disease expression, especially in non-Asian populations. Current research is also exploring the role of inflammation, endothelial dysfunction, and circulating angiogenic factors in disease progression (14; 15).

Despite progress in diagnosis and surgical techniques, challenges remain, particularly in early detection, long-term management, and identifying targeted therapies. Multidisciplinary collaboration is essential for optimizing patient outcomes, especially in complex pediatric cases involving systemic coagulopathy or autoimmune components

## CONCLUSION

Moyamoya disease is a rare but serious cerebrovascular disorder that requires early identification and comprehensive management to prevent long-term neurological deficits. The case presented highlights the importance of considering Moyamoya in children with recurrent neurological symptoms, especially when imaging reveals characteristic vascular patterns. The presence of autoimmune and thrombotic markers should prompt evaluation for Moyamoya syndrome.

Timely surgical revascularization, as demonstrated with STA-MCA bypass, can significantly reduce the risk of recurrent ischemia and improve cerebral perfusion. Long-term prognosis depends on early intervention, effective postoperative care, and regular

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follow-up. Genetic studies and advances in neuroimaging continue to enhance diagnostic accuracy and may eventually lead to personalized therapeutic approaches.

As research into the molecular mechanisms and inflammatory pathways of Moyamoya disease evolves, it holds promise for novel treatment strategies beyond surgical intervention. For now, a multidisciplinary approach remains essential for managing this complex and potentially debilitating condition.

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