Case Report

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Seizure and Stroke in a Patient with Down Syndrome Diagnosed with Moyamoya Syndrome: A Case Report

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ABSTRACT

Moyamoya syndrome (MMS) is a rare, progressive occlusive vasculopathy characterized by stenosis of the distal internal carotid arteries and the formation of collateral vessels resembling a “puff of smoke” on angiography. Patients with Down syndrome (DS) have a significantly higher risk of MMS; however, diagnosis is often delayed due to underlying intellectual disability.

We report the case of a 21-year-old male with Down syndrome who presented with recurrent ischemic strokes. Initial magnetic resonance imaging (MRI) revealed scattered infarcts in both hemispheres. Magnetic resonance angiography (MRA) and digital subtraction angiography (DSA) demonstrated bilateral terminal internal carotid artery stenosis with compensatory anastomoses from the external carotid arteries, confirming the diagnosis of moyamoya syndrome. Due to adequate collateral circulation, the patient was managed conservatively with dual antiplatelet therapy rather than surgical revascularization. Unfortunately, the clinical course was complicated by Stevens–Johnson syndrome following antiseizure medication and subsequently severe pneumonia with bilateral pleural effusion, which ultimately led to the patient’s death.

While confirming previous findings on the diagnostic challenges of MMS in DS, this report emphasizes that systemic complications pose a serious threat to these patients.

Introduction

Moyamoya vasculopathy is a chronic and progressive occlusive condition affecting the distal portions of the internal carotid arteries and the circle of Willis. The resultant ischemia leads to compensatory angiogenesis and the development of a network of collateral vessels, which on angiography resembles a “puff of smoke”—referred to in Japanese as “moyamoya” [1]. Moyamoya syndrome has been reported following various conditions, the most important of which are neurofibromatosis type 1, Down syndrome, thyroid disorders, and sickle cell anemia. The clinical manifestations of MMS include recurrent ischemic strokes, hemodynamic transient ischemic attacks (TIAs), recurrent seizures, and hemorrhage. Various

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other symptoms may also be present, including motor deficits, sensory symptoms, speech disorders, and headache [2]. The aim of this case report is to describe the clinical course of a patient with Down syndrome and moyamoya syndrome, and to raise awareness among clinicians regarding strategies that can be adopted to improve the care of similar patients.

Case Presentation

The patient was a 21-year-old male with Down syndrome who presented with left-sided hemiparesis. Brain MRI (DWI sequence, November 2024) demonstrated scattered acute infarcts in both hemispheres, predominantly involving the cortical regions of the right hemisphere and the right frontal lobe (Figure 1).

Interestingly, initial imaging also revealed evidence of an old, asymptomatic infarction in the right parietal lobe, with no prior history of corresponding neurological symptoms. This incidental finding underscores the diagnostic challenge in patients with Down syndrome, where intellectual disability may mask or delay the recognition of neurological deficits.

The patient was placed on dual antiplatelet therapy. Subsequently, the patient experienced several episodes of seizures, for which levetiracetam was initiated. The patient was discharged from the hospital with prescribed medications. Three months later, the patient returned with new-onset right-sided hemiparesis. Repeat MRI revealed a new infarct at multiple sites within the left insula, left basal ganglia, and left frontal lobe. MRA of the cerebral and cervical vessels demonstrated multiple stenoses, leading to a referral to a neurointerventional fellowship. Digital subtraction angiography (DSA) showed stenosis and occlusion of the bilateral terminal internal carotid arteries, along with anastomosis from the bilateral external carotid arteries (ECAs), consistent with moyamoya syndrome. The characteristic “puff of smoke” appearance was identifiable in the basal ganglia region (Figure 2). DSA (April 2025) confirmed bilateral terminal ICA stenosis and occlusion, with prominent collateral anastomoses from the ECA territory and the classical “puff of smoke” network of moyamoya vessels at the base of the brain (Figure 3). Based on DSA findings, the patient was classified as having moyamoya syndrome at Suzuki stage III–IV. Despite recurrent ischemic strokes, the presence of robust compensatory anastomoses from the bilateral ECAs indicated adequate cerebral perfusion. Therefore, after multidisciplinary discussion, conservative medical management with dual antiplatelet therapy was continued, and revascularization surgery was

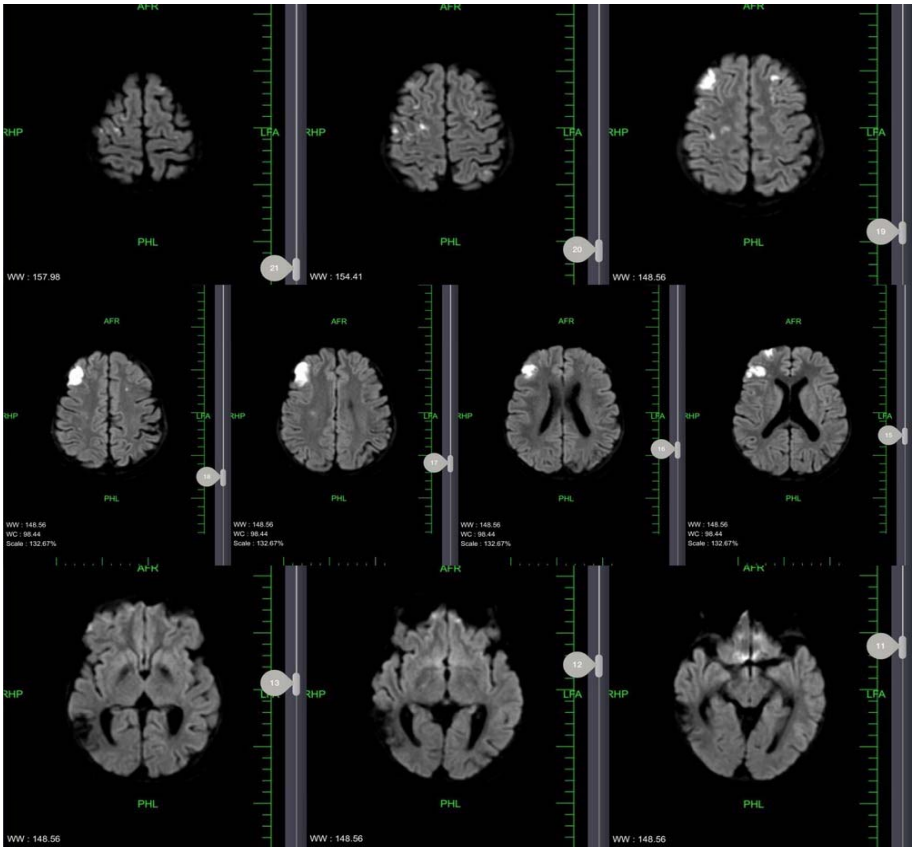

Fig. 1. brain MRI , DWI sequences , november 2024

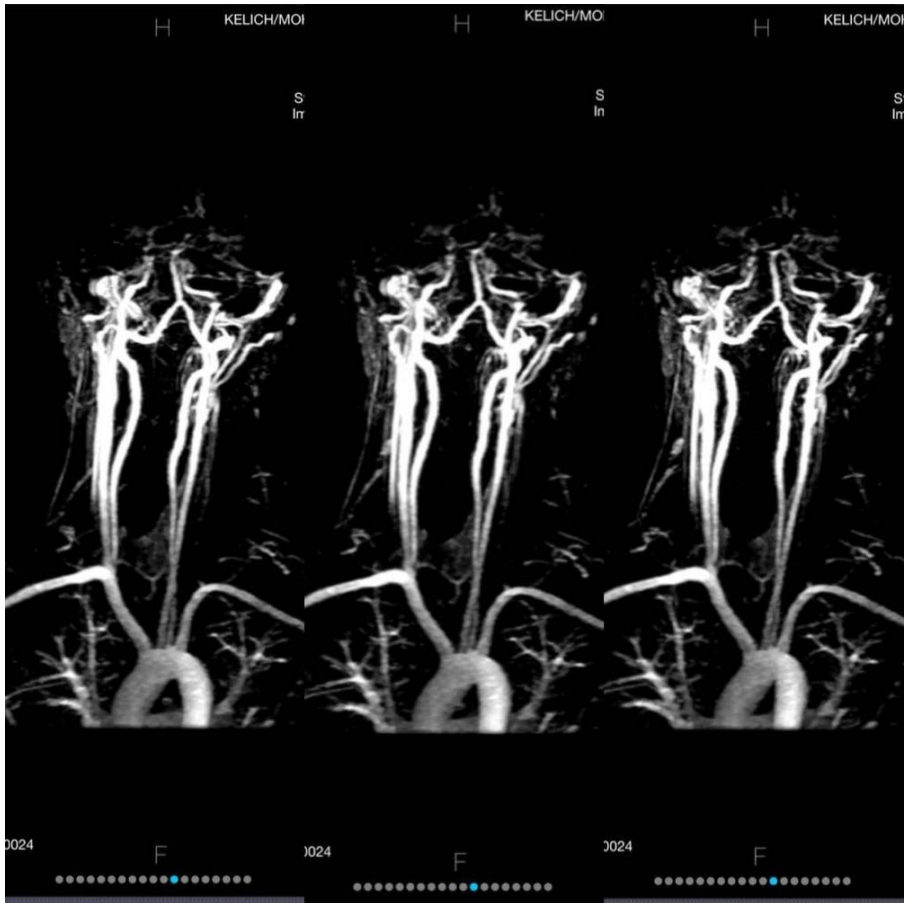

Fig. 2. MRA of brain & neck vessels , April 2025

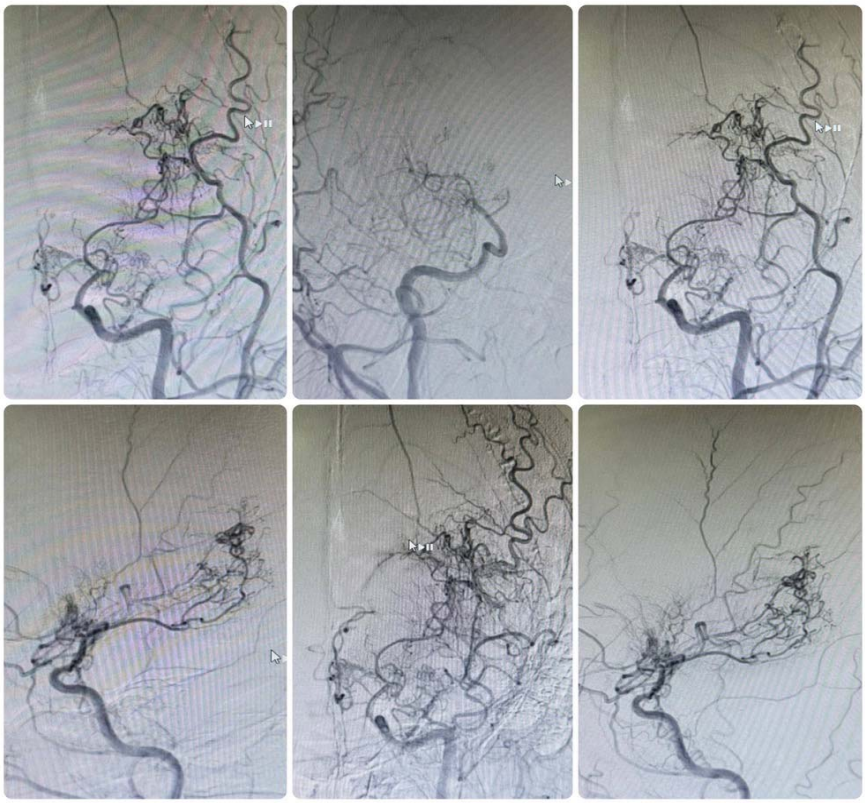

Fig. 3. MRA of brain & neck vessels , April 2025

deferred. This decision was made considering the potential surgical risks and the patient's overall clinical condition.

Two months later, the patient developed Stevens–Johnson syndrome. Following consultation with a dermatologist, treatment with sodium valproate was initiated, and the patient was discharged in good general condition. Unfortunately, one month later, the patient was hospitalized with severe pulmonary manifestations. CT scans of the lungs (July 2025) showed bilateral extensive pneumonia with dense consolidation in both lower lobes and bilateral massive pleural effusion, representing the terminal infectious complications leading to patient demise (Figure 4). Despite all therapeutic interventions, including intubation and placement of a pleural catheter, the patient could not tolerate the disease course and expired. The disease timeline of the patient is reported in Table 1.

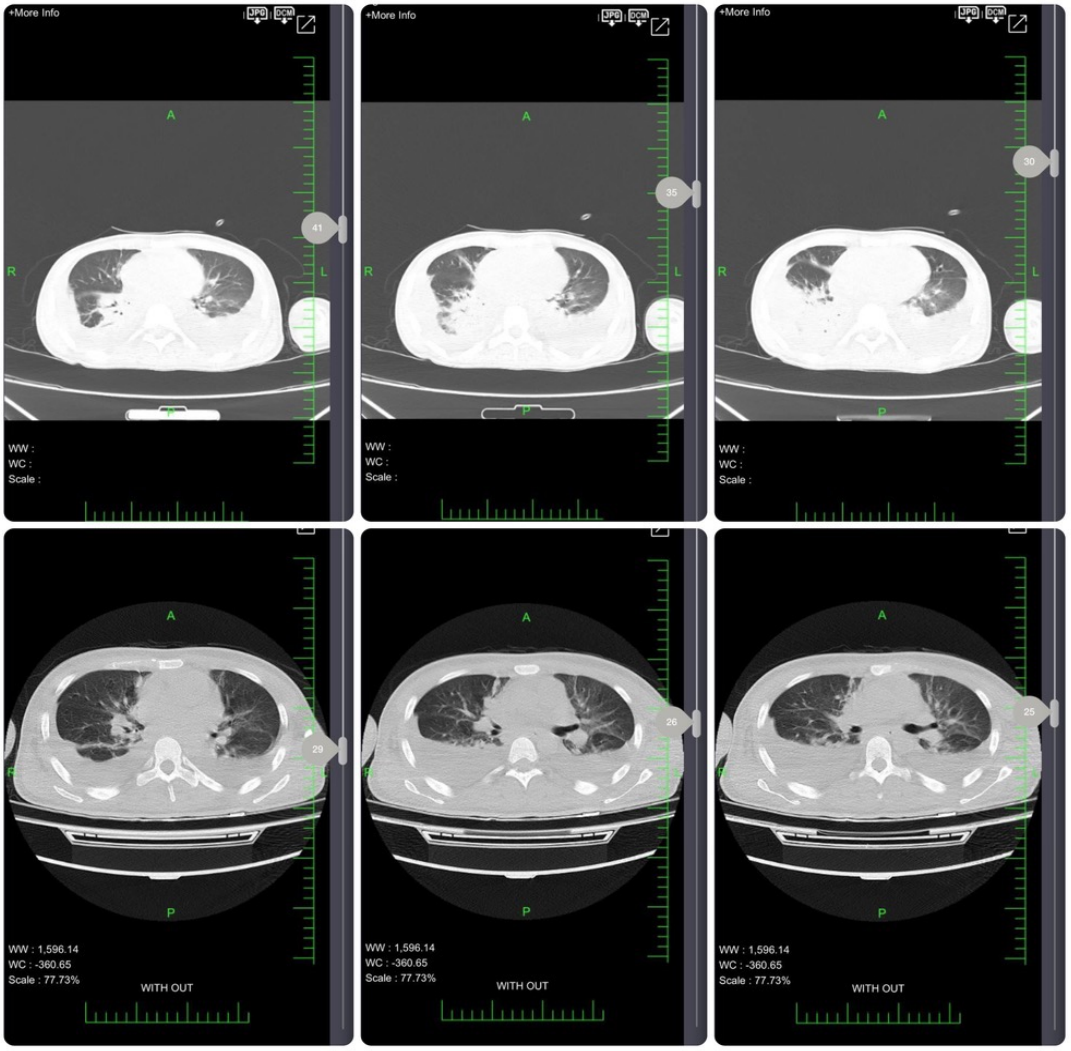

Fig. 4. CT Scans of the Lungs , July 2025

Table 1. Disease Timeline of the Patient

Date	Event	Intervention	Outcome
November 2024	Initial presentation: left-sided hemiparesis	MRI, dual antiplatelet therapy, levetiracetam	Discharged with medications
April 2025	New-onset right-sided hemiparesis, recurrent seizures	Repeat MRI, MRA, DSA; diagnosis of MMS; continued dual antiplatelet therapy	Diagnosis confirmed; no surgery
June 2025	Development of Stevens–Johnson syndrome	Dermatology consult; sodium valproate initiated	Discharged in good condition
July 2025	Hospitalization with severe pneumonia and bilateral pleural effusion	Intubation, pleural catheter, supportive care	Expired

Discussion

In this case report, we describe the clinical course of a 21-year-old patient with Down syndrome (DS) and moyamoya syndrome (MMS), discussing our findings within the context of existing knowledge. The primary objective is to highlight the unique diagnostic and management challenges inherent to this vulnerable population. The imaging findings in our patient, including bilateral terminal internal carotid artery (ICA) stenosis and the characteristic "puff of smoke" appearance, clearly confirm the diagnosis of MMS. These findings are entirely consistent with the report by Shivashankar et al. (2025) in India, who described near-complete bilateral ICA occlusion in a 7-year-old boy with DS [1]. Furthermore, the bilateral vascular involvement in our patient aligns with the findings of Rose et al. (2022), who reported severe bilateral ICA stenosis in an 18-year-old female with DS [3]. Clinically, our patient's presentation with recurrent ischemic strokes followed by seizures represents a relatively common pattern in MMS. Valverde et al. (2024) in Brazil also reported a case of recurrent stroke in a 7-year-old girl presenting with aphasia, motor deficits, and gelastic seizures [4]. This consistency highlights the diversity of neurological manifestations in MMS and underscores the importance of attending to symptoms beyond classic motor weakness.

One of the notable and cautionary findings in our case was the presence of asymptomatic old infarcts on initial imaging. Our patient had evidence of an old infarction in the right parietal lobe with no prior history of neurological symptoms. This phenomenon is clearly consistent with the diagnostic challenge noted by Rose et al. (2022): "Given the near-universal prevalence of intellectual disability in this population, neurological symptoms may be overlooked or lead to delayed diagnosis" [3]. Shivashankar et al. (2025) also emphasized this point, stating that children with comorbid conditions, including DS, require early identification and timely referral to specialized centers [1]. Our finding strengthens the argument that periodic neurovascular screening, even in patients without overt neurological symptoms, may be necessary in this population.

An important differential point between our study and previous studies was observed in the therapeutic approach. In our patient, due to the presence of adequate compensatory anastomoses from the external carotid artery (ECA) on angiography, the need for revascularization surgery was not indicated, and treatment with dual antiplatelet therapy (aspirin) was continued. This stands in contrast to Rose et al. (2022), who performed indirect revascularization surgery (EDAS) on their patient [3], and to the study by Shivashankar et al. (2025), in which treatment was aimed at surgical cerebral revascularization [1]. This difference emphasizes that the choice of therapeutic approach in MMS should not be uniform, but rather should be decided based on individual factors such as age, symptom severity, extent of vascular involvement, and most importantly, the presence of sufficient compensatory collateral vessels. In some cases, conservative medical therapy may be as effective as surgery.

Unfortunately, the clinical course of our patient was complicated by catastrophic adverse events, including Stevens–Johnson syndrome (following the use of antiseizure medications) and subsequently severe pneumonia with bilateral pleural effusion, which ultimately led to the patient's death. This finding represents a critical point of divergence between the present report and previous studies. While Rose et al. (2022) noted an increased risk of congenital heart disease in DS [3], none of the reviewed studies [1,3,4] specifically addressed the high risk of medication-related complications (such as severe cutaneous reactions) and infectious complications (such as severe pneumonia) in these patients. Patients with DS are an extremely vulnerable population due to immune dysregulation and potential drug sensitivities. Our report serves as a serious warning in this regard, demonstrating that mortality in these patients may arise not only from MMS itself and its associated strokes but equally from treatment-related adverse effects and intercurrent infections.

As a case report, this study inherently has limitations in generalizability. Nevertheless, its qualitative findings are highly valuable. As Rose et al. (2022) emphasized the necessity of advanced imaging, immunological, and epigenetic studies [3], we likewise believe that understanding the underlying mechanisms of the DS–MMS association requires further research. Specifically, future studies should focus on developing standardized screening protocols for asymptomatic old infarcts, as well as clinical guidelines for the prevention and management of medication-related and infectious complications in this vulnerable population.

Seizures in moyamoya syndrome are thought to arise from cortical irritation secondary to recurrent ischemia, particularly when infarcts involve the frontal and insular regions. In our patient, the development of seizures following recurrent strokes was managed with levetiracetam, which proved effective in seizure control. No other identifiable cause of seizures—such as metabolic disturbances or intracranial infection—was identified.

Conclusions

In summary, while confirming the findings of previous

studies regarding the diagnostic challenges of MMS in patients with DS, the present report emphasizes three novel and essential points: (1) the presence of asymptomatic old infarcts underscores the necessity of periodic neurovascular screening; (2) the presence of compensatory anastomoses from the ECA may obviate the need for surgery in some patients; and (3) most importantly, systemic complications (medication-related and infectious) represent a serious and potentially fatal threat to these patients. Therefore, the management of these patients requires a comprehensive, multidisciplinary approach involving specialists in neurology, neuroradiology, neurosurgery, infectious diseases, dermatology, and intensive care. The death of our patient serves as a somber warning that the treatment of MMS in Down syndrome extends beyond cerebral revascularization and must take into account the entire immune system and physiology of these patients. Further studies with larger sample sizes are an undeniable necessity to develop specific diagnostic and therapeutic protocols for this vulnerable population.

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Ethical Considerations

Compliance with ethical guidelines

There were no ethical considerations to be considered in this article.

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Conflict of Interests

The authors declare that they have no conflict of interest.

Ethics approval

Ethics approval and consent to participate (SEMUMS, Ethical code: IR. SEMUMS.REC. 1404.319)

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