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Case Report

Moyamoya disease as a cause of intracranial haemorrhage in an infant: a case report

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ABSTRACT

Moyamoya is a rare cerebrovascular condition characterized by progressive narrowing of large intracranial arteries leading to development of prominent arterial collaterals. These collateral vessels on angiography produce a smoky appearance termed as "moyamoya", a Japanese word meaning puff of smoke in the air. Moyamoya disease usually presents as ischemic stroke in children and hemorrhagic stroke in adults. We present the story of an infant, term appropriate for gestational age with meconium aspiration syndrome, vigorous with hypoxic ischemic encephalopathy stage 1 (Sarnat with focal seizures involving right side of the body and severe anaemia, who was brought to our ER with complaints of vomiting and abnormal movements involving right side of the body. On examination severe pallor was observed, accompanied by a bulging anterior fontanelle, CT angiogram demonstrated marked attenuated calibre of left internal carotid artery in the supra-clinoid part A1 segment of anterior cerebral artery and middle cerebral artery with subacute infarct in middle cerebral artery territory suggestive of Moyamoya Disease. The infant was managed conservatively with one aliquot of packed red blood cells and antiepileptic drug phenobarbital. As our center is not equipped with neurosurgical interventions to be done in such patients, baby was referred to higher center for surgery. This case report serves as a reminder to consider Moyamoya disease in the differential diagnosis of infantile cerebral haemorrhage (stroke). Early diagnosis and prompt management are essential for improving outcomes in this rare and potentially devastating condition.

Keywords: Moyamoya disease, Moyamoya syndrome, Rare disease, Stroke, Direct revascularization, Indirect revascularization

INTRODUCTION

Moyamoya is a progressive steno-occlusive disease typically involving the distal internal carotid artery (ICA), proximal MCA bilaterally, and not infrequently the anterior cerebral arteries (ACA). This arterial steno-occlusive disease is associated with development of extensive collaterals at the base of the brain, most commonly enlargement of lenticulostriate vessels producing the "moyamoya" (Japanese for "puff of smoke"). Moyamoya has a bimodal incidence with a peak at 5 years and a smaller peak in the fourth decade.

Moyamoya usually presents with bland ischemic stroke in childhood, whereas adults frequently present with haemorrhagic stroke. Moyamoya is often not diagnosed until stroke occurs.

Acute stroke heralds the diagnosis of moyamoya in approximately half of all children with the disease, despite a history of transient ischemic attacks (TIAs) in many patients.[1,2] Children with moyamoya may present with headache and TIAs. TIAs frequently precede a sentinel stroke, which is confirmed by imaging. Signs of anterior circulation ischemia like aphasia, dysarthria,

hemiparesis, and seizures are seen often, and less commonly presents as, syncope, visual changes, and chorea. Symptoms are often provoked by hyperventilation, for example with crying or exercise, as a result of hypocarbia-induced vasoconstriction. Moyamoya may be idiopathic ("moyamoya disease") or occur in association with syndromes ("moyamoya syndrome"), such as sickle cell disease and trisomy 21 or as the sequela of injury.

Moyamoya is associated with many underlying genetic disorders, including sickle cell disease, trisomy 21, Noonan syndrome and neurofibromatosis type 1.[3-5] Moyamoya disease is likely a genetic disease, most probably polygenic, based on the high prevalence of moyamoya in Asia and familial aggregation in 5% to 10% of cases. Approximately 15% of patients with non-syndromic moyamoya (moyamoya disease) have a family history of moyamoya.

The mode of inheritance appears to be autosomal dominant with incomplete penetrance. Familial moyamoya in the Asian population has been linked to a susceptibility region on 17.q25, a ring finger domain containing protein 213 or RNF213.[3,6-8] Given the high risk of stroke in moyamoya, revascularization surgery is recommended for symptomatic patients to decrease the risk of further TIAs and ischemic and hemorrhagic stroke, as well as for asymptomatic patients who are at risk of progression. Revascularization can be performed by direct or indirect measures.

Direct revascularization most commonly employs anastomosing the superficial temporal artery to the middle cerebral artery. Indirect revascularization usually involves pial synangiosis where the superficial temporal artery is placed on the surface of the brain. Direct revascularization has the advantage of immediate improvement in flow but is technically demanding, requiring a minimal artery size and therefore is not an option in very young children.[9]

Indirect revascularization has the advantage of not being limited to a particular arterial territory but requires months to show benefit as collaterals take time to develop. In children, direct and indirect revascularization appears equally efficacious with most centers preferring indirect revascularization due to lower surgical morbidity. Revascularization not only reduces the incidence of ischemic stroke in childhood, but appears to decrease the risk of haemorrhagic stroke in adulthood as well.[10] Revascularization should be pursued promptly in eligible children, particularly very young children, in whom the disease can be more aggressive with poor outcome.

This report presents a rare case of haemorrhagic moyamoya disease in a 30-days old infant with a complex medical history. Moyamoya, characterized by progressive bilateral stenosis of the intracranial arteries, is uncommon in paediatric population, especially infants, making this case particularly unique and challenging.

CASE REPORT

A 30-day old male neonate, fourth in birth order, born via normal vaginal delivery, term appropriate for gestational age, with meconium aspiration syndrome at birth requiring resuscitation with hypoxic-ischemic encephalopathy (HIE) stage 1, presented with complains of abnormal movements involving the right side of the body and vomiting.

On examination severe pallor was observed, accompanied by a bulging anterior fontanelle. Blood pressure was within normal range and fundus examination revealed no abnormality. CNS examination was normal. Rest of the systemic examination was within normal range.

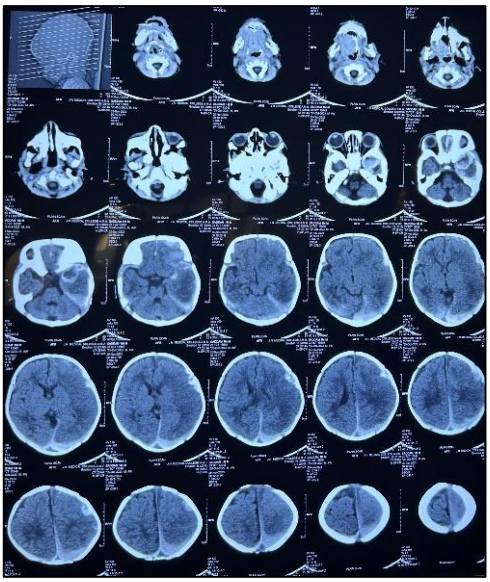

Figure 1: CT scan of head.

Figure 1 is showing, intra axial hyper density with CT value of 20-30 HU suggestive of intraparenchymal bleed involving left temporal lobe (size 19×23×21mm) with a hypodense fluid noted within it forming fluid-fluid level, extra axial hyperintensity with CT value of blood suggestive of extra axial bleed overlying left cerebral hemisphere and right parieto-temporo-occipital lobe (maximum thickness 11 mm), extra axial bleed along falx and bilateral tentorium cerebelli.

On computed tomography of head (Figure1), findings consistent with intraparenchymal bleed involving left temporal lobe and extra axial bleed overlying left cerebral hemisphere and right parieto-temporo-occipital lobe and also along falx cerebri and bilateral tentorium cerebelli was present. Laboratory investigations showed severe anaemia with haemoglobin level of 3.1 g/dl, while

platelet count and coagulation profile were normal. Peripheral blood smear revealed no signs of haemolysis.

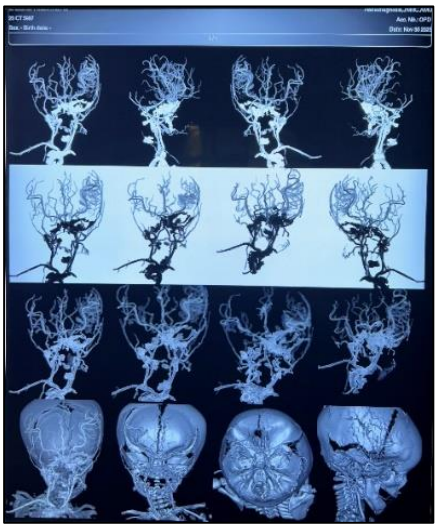

Figure 2: CT scan of Angiogram.

Figure 2 is showing marked attenuated calibre of left internal carotid artery in the supraclinoid part, A1 segment of anterior cerebral artery and middle cerebral artery and its branches with gyral enhancement.

Thereafter, CT angiogram (Figure 2) was done which demonstrated marked attenuated calibre of left internal carotid artery in the supra-clinoid part A1 segment of anterior cerebral artery and middle cerebral artery with subacute infarction suggestive of Moyamoya. Baby was managed conservatively with one aliquot of packed red blood cells and antiepileptic drug phenobarbital. As our center is not equipped with neurosurgical interventions to be done in such patients, baby was referred to higher center for surgery.

DISCUSSION

Why is this disease rare?

WHO defines rare disease as often debilitating lifelong disease or disorder with a prevalence of 1 or less, per 1000 population 11. There are no community-based studies from India. In Japanese epidemiologic surveys, prevalence of MMD is 3.2-10.5/100,000 population. This report presents a yet rarer form of haemorrhagic moyamoya disease in a 30-days-old infant. Moyamoya usually presents with ischemic stroke in childhood, whereas adults frequently present with haemorrhagic stroke. Moyamoya is uncommon in paediatric population, especially infants, making this case particularly unique and challenging.

This case highlights the potential for moyamoya disease to present with early-onset haemorrhagic complications in a neonate with additional complex medical conditions. CT angiogram findings confirm the presence of moyamoya disease. Amlie-Lefond et al reported a case of moyamoya in 2 months 3 weeks old infant who presented with seizures and ischemic stroke.[12] The management of such a case requires a multidisciplinary approach, including neonatology, paediatric neurology, and neurosurgery. Supportive care for anaemia and seizure control is crucial, along with close monitoring for further neurological complications. The role of revascularization surgery in this specific case needs careful consideration.[13,14]

CONCLUSION

This case report serves as a reminder to consider moyamoya disease in the differential diagnosis of neonatal cerebral haemorrhage, even in the presence of other potential contributing factors. Early diagnosis and prompt management are essential for improving outcomes in this rare and potentially devastating condition.

Recommendations

Genetic testing for known moyamoya-associated mutations could be explored to assess familial risk and potential future recurrence. Regular neuroimaging follow-up is crucial to monitor disease progression and potential complications. Multidisciplinary discussions involving neonatology, paediatric neurology, and neurosurgery are essential for developing an optimal management plan for this complex case.

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