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Case Report

Hyperacute anterior spinal artery infarction in a child with prothrombin G20210A mutation: a case report with brief review of literature

Sapna Singh, Praveen Kumar*, Rama K. Sabharwal, Divya Agrawal

Department of Paediatric Neurology, Institute of Child Health, Sir Ganga Ram Hospital, New Delhi, India

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***Correspondence:**
Dr. Praveen Kumar,
E-mail: drpraveenk@icloud.com

ABSTRACT

Anterior spinal artery territory infarction in children is rare, resulting from hypoperfusion or occlusion of the anterior spinal artery. The ischemia involves the anterior two-thirds of the spinal cord. It presents acutely with pain, para- or quadriplegia, dissociated sensory loss, bowel and bladder involvement, respiratory distress, and autonomic dysfunction. We present a case of a 14-year-old previously healthy girl, who presented with hyperacute quadriplegia and respiratory failure. Physical examination was compatible with a high cervical spinal cord lesion. Magnetic resonance imaging (MRI) of the spine with diffusion weighted imaging (DWI) revealed lesion of the ventral cervical cord between C2 and C6 levels, suggestive of infarct in the anterior spinal artery territory. Genetic studies revealed a prothrombin G20210A heterozygous mutation. She was treated with anticoagulants and supportive care. She showed good improvement and was ambulant 6 months later.

Keywords: Spinal cord infarction, Quadriplegia, Prothrombin G20210A

INTRODUCTION

Spinal cord infarction (SCI) is rare in children, and typically occurs as rapid onset, painful acute myelopathy. Prompt etiological diagnosis is important for immediate and long-term management and prognostication. SCI is often misdiagnosed as transverse myelitis.[1,2] Causes of hyperacute (<6 hours) or acute (12–48 hours) paraplegia/quadriplegia with sensory loss and bowel/bladder dysfunction are limited, typically including spinal cord infarction or hemorrhage, trauma, transverse or viral myelitis, and acute cord compression.

The study group on spinal cord infarction of the French Neurovascular Society identified 28 patients with SCI from 16 centers over two years (almost one patient per hospital per year).[3] Other centers reported 24-27 patients seen over a 14-year period.[4,5] Prothrombotic states should be considered in children in cryptogenic cases or those with family history of thrombosis. The prothrombin G20210A mutation is a known risk factor, increasing the risk of deep vein thrombosis, stroke, and fetal loss.

CASE REPORT

A 14-year-old girl with unremarkable birth and development history, presented with acute neck and shoulder pain followed by inability to stand, and developed weakness of all extremities. She was admitted to a local hospital; and subsequently developed breathing difficulty and required mechanical ventilation. There was no associated headache, vision changes, seizure, or loss of consciousness, no history of trauma, lifting weights, fever, or drug ingestion. Magnetic resonance imaging (MRI) brain was normal, and MRI spine was suggestive of demyelination in the C2 to C6 segments. Lumbar puncture revealed a normal cerebrospinal fluid (CSF) analysis. She received an IV pulse dose of methylprednisolone, and 2 gm/kg of IVIg over 5 days. As there was no significant improvement child was shifted to our hospital.

On examination she was conscious on ventilatory support with stable vitals. There was flaccid quadriplegia, generalized areflexia and mute plantar responses bilaterally. Sensations could not be accurately judged in view of her critical condition. Bladder and bowel sensations were absent.

Despite acute SCI being the most probable diagnosis, a thorough investigation was conducted to exclude other causes. Nerve conduction study was normal. Contrast MRI spine showed prominent "pencil-like" hyper intensities on DWI sequence involving the ventral cord parenchyma from C2-C6 vertebral body (Figure 1). Corresponding low apparent diffusion coefficient (ADC) image was seen (Figure 2); there were no signal abnormalities in the vertebrae or discs. Further testing, including hemogram, sickling test, blood biochemistries, electrolytes, prothrombin time, partial thromboplastin time, prothrombotic markers, serology for systemic vasculitis were normal.

Figure 1: DW1 images, well defined restricted diffusivity seen in the ventral cervical cord parenchyma.

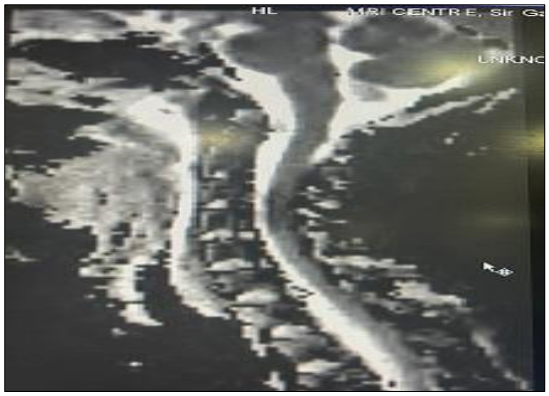

Figure 2: Corresponding low ADC values visualized opposite to C3 and C4 vertebrae.

Homocysteine, antithrombin 3, protein C and S, antiphospholipid antibodies panel revealed no abnormalities. Heterozygous mutation of prothrombin G 20210 was positive. Electrocardiogram and echocardiography were normal. The child was anticoagulated with enoxaparin. Supportive care and physiotherapy were started. She needed tracheostomy and spent the next 6 weeks in intensive inpatient rehabilitation, with gradual improvement in the movement of her right arm. At the time of discharge, she was able to accept orally, had gained bladder/bowel control and started drawing with her right hand. Five months after discharge, she was walking with support. One year later, she had independent walking but had grade 3/5 power in her arms.

DISCUSSION

The spinal cord is supplied by 1 anterior and 2 posterior spinal arteries, which extend longitudinally to conus medullaris. The arteries are connected by pial plexus surrounding the spinal cord. The circulation to spinal cord has a rich anastomotic anatomy of the cord that results in rarity of spinal cord infarction in comparison to cerebral infarction. The peak prevalence of SCI is between the sixth and seventh decades of life and is rare in pediatric age group.[6]

SCI presents acutely with pain, paralysis below the lesion, loss of pain and temperature sensation with preserved vibratory and position sense, absent deep tendon reflexes in acute phase, autonomic dysfunction, and sphincter loss. Clinical features vary, ranging from paraplegia/ quadriplegia to mild weakness. Incomplete forms include anterior horn syndrome (flaccid paralysis, areflexia), Brown-Séquard syndrome, and "man-in-the-barrel" syndrome from central cord involvement. Infarcts most commonly affect the lower thoracic cord; cervical involvement is rarer but more severe in children.

In children, diverse aetiologies for SCI include vasculitis, severe hypotension, fibrocartilaginous embolism, congenital AV malformations, meningitis, thrombophilia, primary antiphospholipid syndrome, congenital cardiovascular anomalies and trauma-induced vasospasm. A trigger such as a fall, gymnastics, neck flexion/ extension, Valsalva manoeuvre is found in up to 48% of cases. In children, the relatively less elastic spinal cord is strained within a more flexible spinal column; vasospasm can arise from hyperflexion injuries, which may be complicated by poor spinal cord pressure autoregulation leading to ischemia.[7] Local arteriovenous shunting may reduce perfusion, causing localized infarction. In neonates, umbilical artery catheters may disrupt the artery of Adamkiewicz, leading to thoracic cord infarction.

MRI of spinal cord is the investigation of choice, aiding diagnosis, and ruling out myelitis or cord compression. Although DWI and ADC are not routinely performed, they are sensitive for spinal ischemia. SCI can be detected by DWI within 3-4 hours, while T2-weighted imaging may take 24 hours or more. MRI may appear in up to 24% patients if done in first 24 hours.[2] Detection of vertebral body T2 hyperintensity on MRI is a strong indicator of SCI. "Pencil- like" hyperintensities in the ventral cord on sagittal DWI (Figure 1) or T2 weighted images; and the

classical "owl's eye sign" involving central- anterior cord substance is seen in axial images (Figure 2). Thus, DWI should be routinely performed in acute myelopathies.

Meta-analyses confirm that genetic mutations contribute to both arterial and venous infarcts, regardless of mutation type or variability. Prothrombotic abnormalities may arise from defects in coagulation, fibrinolysis, endothelium, or platelets. Combined genetic defects carry a higher thrombosis risk than single-gene mutations. The prothrombin G20210A mutation is a prevalent risk factor for thrombosis, which elevates the likelihood of deep vein thrombosis, stroke, and foetal loss.[8-12] Prothrombin (PT) G20210A is one of the genetic polymorphisms associated with thrombophilia, and is the second most common mutation after factor V. This mutation results in unchecked thrombin formation predisposing to the thromboembolic phenomenon. Young et al found G20210 A mutation in 38 out of 374 children, from 26 centres, who suffered a thrombotic event (arterial or venous).

Thirty-six patients were heterozygotes and two homozygotes. The mean age of all the patients was 7.6 years. Association with infarcts of the spinal cord and with cerebral sinus thrombosis was noted.[11] Prothrombin G20210 mutation has been reported in spinal cord infarction in children.[8-13] Studies show this polymorphism is rare in Africans and Asians, and has been rarely reported from India. However, Salomi et al found the PT G20210A is prevalent in Indian patients, with cryptogenic arterial or venous strokes, especially among those from north India.

CONCLUSION

In conclusion, SCI is a rare cause of quadriparesis or paraparesis in children. A sudden onset with pain and paralysis, along with sensory, autonomic, and sphincter involvement, strongly suggests the diagnosis. DWI is essential for early detection, as T2-weighted MRI may miss lesions. Thrombotic workup is crucial, as treatable thrombophilic conditions may require long-term anticoagulation, especially in posterior circulation strokes. Addressing physical and psychosocial complications and initiating early physiotherapy and occupational therapy can improve outcomes. There are no standardized treatment guidelines; therapies used include antiplatelets, anticoagulants, IVIG, steroids, and plasmapheresis, especially when transverse myelitis is misdiagnosed.

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