Case Report

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Deep Medullary Vein Thrombosis in a Term Neonate

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ABSTRACT

Introduction: Deep medullary vein thrombosis (DMVT) is a rare but significant cause of neonatal stroke, characterized by venous congestion and thrombosis in subcortical white matter. Early identification is crucial due to its association with long-term neurological sequelae.

Case Description: We report the case of a term neonate presenting with focal seizures at 48 hours of life. Prenatal history was significant for fetal growth restriction and vacuum-assisted delivery. Neuroimaging revealed radial, fan-shaped T2 hypointense lesions in periventricular white matter as hallmark findings of DMVT. Compound heterozygosity for MTHFR mutations was identified. Seizure management with phenobarbital and supportive care were prioritized, with serial imaging showing favorable evolution. At 12 months, the child exhibited an unremarkable neurologic examination and appropriate psychomotor development without recurrent seizures.

Discussion/Conclusion: This case highlights the importance of early neuroimaging in neonates with seizures or atypical neurological presentations. Enhanced awareness and understanding of DMVT among clinicians may reduce diagnostic delays, ultimately improving outcomes. The absence of standardized treatment protocols underscores the need for further research.

INTRODUCTION

Deep medullary vein thrombosis (DMVT) is a rare but increasingly recognized cause of neonatal stroke, both in preterm and term infants.(1,2) The deep medullary veins, integral to the venous drainage of subcortical white matter, are vulnerable to congestion and thrombosis. Neonates, particularly those experiencing perinatal complications such as asphyxia, infections, or prothrombotic conditions, are at heightened risk of DMVT.(2)

Recent advancements in neuroimaging, particularly the use of MRI, have facilitated the identification of characteristic patterns of DMVT, such as fan-shaped linear T2 hypointense lesions in periventricular white matter.(1–3) These lesions are often accompanied by ischemic injury or hemorrhagic infarction.(2) Despite its rarity, DMVT carries a significant risk of long-term neurological sequelae, including epilepsy, motor disabilities, and cognitive impairments, highlighting the need for timely recognition and management.(3)

This case report describes the clinical presentation, diagnostic approach, and management of a term neonate with neonatal seizures associated with deep medullary vein thrombosis, contributing to the growing understanding of this under-recognized condition.

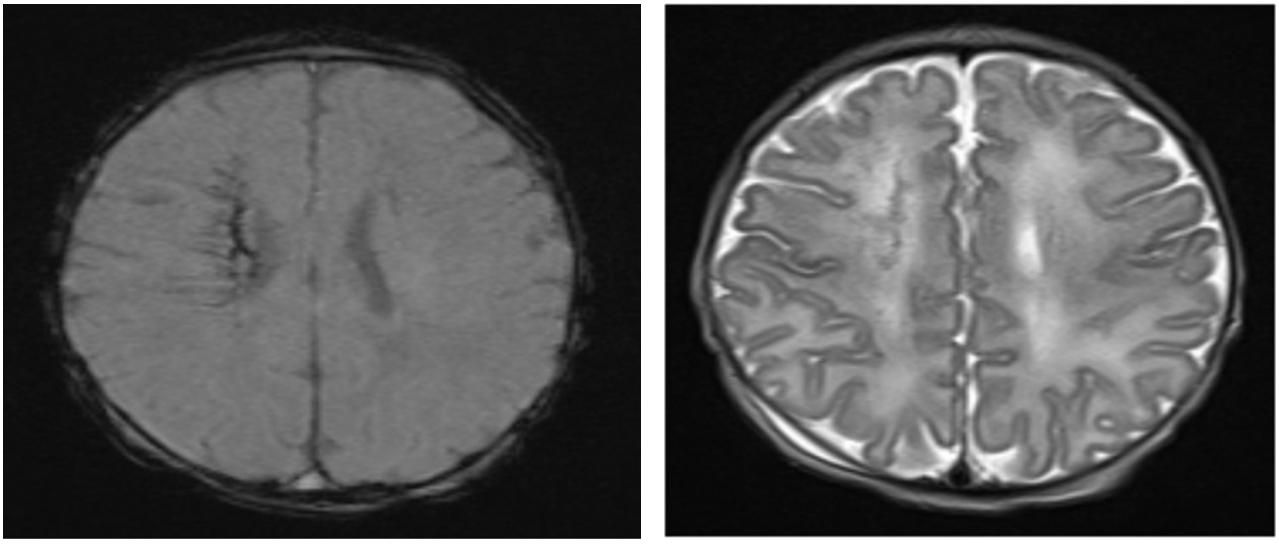
CASE DESCRIPTION

A male term neonate with prenatal history of fetal growth restriction diagnosed in the third trimester and complicated delivery by vacuum extraction, presented at 48 hours of life, with an episode of clonic movements of the left upper limb lasting 20–30 seconds, unresponsive to touch. Additionally, there were reports of feeding difficulties. On clinical examination, the newborn was hemodynamically stable, normothermic, normoglycemic, hypoactive but reactive, with an asymmetric resting posture. The remainder of the physical examination was unremarkable. The infant was admitted to

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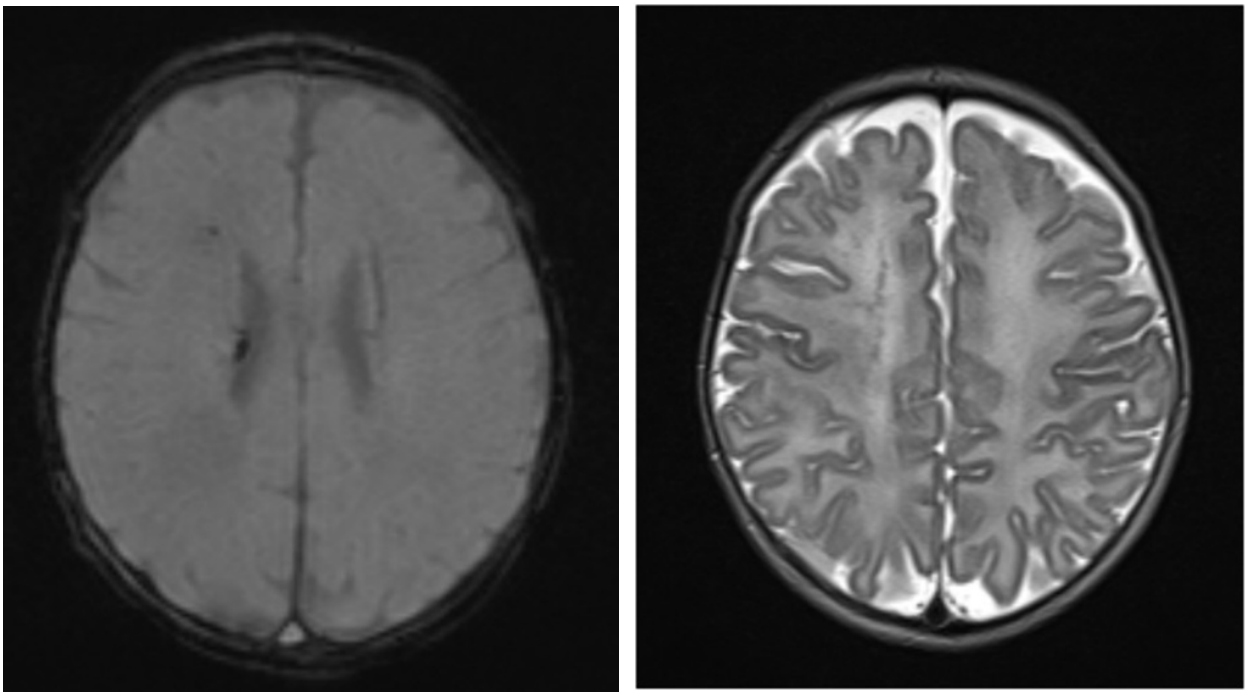
the neonatal intensive care unit (NICU), where a new episode of clonic movements involving the left upper and lower limbs was observed. An initial dose of phenobarbital (20 mg/kg) was administered, and amplitude-integrated electroencephalography (aEEG) monitoring was initiated. To investigate the cause of neonatal seizures, an analytical study, blood cultures, metabolic screening and transfontanellar ultrasound were performed, all yielding normal results. During aEEG monitoring, a new episode of clonic movements of the left upper limb was observed, with corresponding/matching electrographic changes. Phenobarbital was readministered at 10 mg/kg, followed by maintenance therapy at 5 mg/kg/day. Brain MRI, performed on day 3 of life showed deep medullary vein thrombosis (Figure 1). Associated findings included ischemic foci in the right periventricular parenchyma and microhemorrhages in the head of the left caudate nucleus and right basal ganglia. Supportive management was opted for, and anticoagulation therapy was not initiated. Etiological investigation of the stroke was carried out. Electrocardiogram, echocardiogram, and abdominal and renal doppler ultrasounds, were performed, all without abnormalities. Thrombophilia screening in the neonate revealed compound heterozygosity for MTHFR gene, with the C677T and A1298C mutations. Maternal and paternal thrombophilia screening was normal. On day 10, a repeat transfontanellar ultrasound showed ischemic foci consistent with the previous MRI findings, with no new abnormalities. A follow-up brain MRI on day 17 demonstrated favorable imaging evolution (Figure 2). The neonate did not present further clinical or electrographic seizures and showed gradual improvement in his neurological examination. At discharge, the neonate was referred for follow-up in neonatology, pediatric neurology, ophthalmology, otolaryngology, and physical and rehabilitation medicine clinics. At 3 months of life, additional thrombophilia screening was normal, and the EEG remained free of epileptiform activity. Phenobarbital was tapered off and discontinued at 4 months of age. At the current age of 12 months, the child maintains excellent clinical progress, without visible neurological sequelae, appropriate psychomotor development and no seizure recurrence.

Figure 1. Axial susceptibility-weighted (A) and T2- weighted (B) images show small dilated vessels, compatible with deep medullary vein distribution, in the right periventricular white matter (black arrows), with associated edema (B).



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Figure 2. Axial susceptibility-weighted (A) and T2- weighted (B) images demonstrate involution of the hypointense linear aspects in justa-ventricular images of right predominance, documented in the previous examination. This involution suggests deep venous recanalization and resorption of the existing edema.



DISCUSSION

The diagnosis of DMVT in neonates remains challenging due to its non-specific clinical presentation and overlap with other causes of neonatal seizures.(3) In this case, the neonate's initial focal seizures prompted an urgent and comprehensive workup, leading to a early MRI, which revealed the hallmark imaging findings of DMVT: radial, fan-shaped T2 hypointense lesions in the periventricular white matter, coupled with evidence of ischemia and microhemorrhages. These findings align with patterns described in the literature, where DMVT predominantly affects frontal and parietal regions due to the drainage architecture of the deep medullary veins.(1,3)

The pathophysiology of DMVT in neonates often involves a combination of perinatal risk factors such as instrumental delivery, as seen in this case.(2,3) The presence of compound heterozygosity for MTHFR gene mutations (C677T and A1298C) in the neonate may have predisposed to the thrombotic event. However, the clinical relevance of these mutations, remains controversial, underscoring the complexity of interpreting thrombophilia results in neonates.(4)

Treatment strategies for DMVT are not standardized, with no clinical trials or evidence-based guidelines specific to neonates. (3) Anticoagulation therapy, often considered in venous thrombosis,(5,6) was not initiated in this case due to the absence of associated sinus venous thrombosis. Instead, supportive care and seizure management with phenobarbital were prioritized, with earlyimaging reassessment. As favorable imaging evolution was demonstrated, with resolution of acute lesions, the decision of the conservative approach was consolidated.

The neonate's clinical improvement, absence of recurrent seizures, and appropriate neurodevelopmental progress at 12 months highlights the importance of early seizure control, ongoing neurological monitoring, and multidisciplinary follow-up. However, the long-term risk of neurodevelopmental impairment remains a concern.(3) Continued multidisciplinary follow-up is essential to monitor for such outcomes and provide early intervention if necessary.

CONCLUSION

This case highlights the importance of early neuroimaging in neonates with seizures or atypical neurological presentations. Enhanced awareness and understanding of DMVT among clinicians may reduce diagnostic delays, ultimately improving outcomes. Future research should aim to establish evidence-based protocols for the diagnosis and management of this rare but impactful condition.

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