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Case Report

MRI Diagnosis of Lissencephaly in a Neonate with Neonatal Jaundice: A Case Study in Ghana

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Abstract: Lissencephaly is a rare and severe neurodevelopmental disorder caused by defective neuronal migration during the first trimester of embryogenesis, resulting in a smooth cerebral cortex lacking normal gyri and sulci. We present case report of a female neonate, less than one month old who was admitted to NICU at the Komfo Anokye Teaching Hospital in Kumasi, Ghana, presenting with neonatal jaundice and an elongated cranial shape. Further investigations, with a Magnetic Resonance Imaging (MRI) on a Toshiba Achieva 1.5 Tesla scanner, revealed bilateral frontoparietal and temporal lissencephaly, absence of the septum pellucidum, basal ganglia fusion with poor internal capsule definition, and ventricular dilation. These findings were consistent with lissencephaly, likely resulting from tubulinopathy, coexisting with lobar holoprosencephaly. A multidisciplinary approach was employed in the management of this neonate. This case highlights the importance of MRI in diagnosing and understanding complex congenital brain anomalies, particularly in resource-limited settings like Ghana.

Keywords: Lissencephaly, Neuronal migration disorder, Holoprosencephaly, MRI, Tubulinopathy, Basal ganglia fusion.

INTRODUCTION

Neonatal jaundice is a common condition affecting newborns worldwide, resulting from elevated bilirubin levels. If untreated, severe hyperbilirubinemia can lead to kernicterus, a form of chronic bilirubin encephalopathy, characterized by irreversible neurological damage (Shapiro, 2010; Bhutani *et al*., 2004). Effective treatment, such as phototherapy, has been shown to significantly reduce bilirubin levels and prevent long-term complications (Maisels & McDonagh, 2008). Kernicterus remains a global concern, particularly in low-resource settings where timely intervention may be limited (Russ *et al*., 2021).

Concurrent with neonatal jaundice, congenital brain abnormalities such as lissencephaly can complicate the clinical presentation and management. Lissencephaly, a rare malformation of cortical development, arises due to impaired neuronal migration during embryogenesis, leading to a smooth brain surface with absent or reduced gyri (Barkovich *et al*., 2012; Guerrini & Dobyns, 2014). This condition is associated with severe neurological deficits, including developmental delay, seizures, hypotonia, and failure to thrive (Di Donato *et al*., 2017; Hayward *et al*., 1991). Genetic factors, congenital infections such as cytomegalovirus (CMV), and other etiologies have been implicated in the development of lissencephaly (Elias, 2003; Averill *et al*., 2015).

Cytomegalovirus infection, in particular, has been extensively linked to cortical malformations such as lissencephaly and pachygyria. Pre- and postnatal brain imaging of CMV-infected neonates often reveals structural abnormalities such as white matter changes, ventricular enlargement, and cortical malformations (Vanbuggenhout *et al*., 2022; Gowda *et al*., 2021). Magnetic Resonance Imaging (MRI) remains the gold standard for diagnosing these abnormalities, offering high-resolution images that delineate cortical structure and detect subtle anomalies (Shroff *et al*., 2010; Hüppi & Barnes, 1997). MRI findings in congenital CMV infection and lissencephaly often reveal overlapping features, making detailed imaging and clinical correlation essential for accurate diagnosis (Averill *et al*., 2015; Benoist & Ville, 2007).

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Additionally, cranial abnormalities such as craniosynostosis, the premature fusion of cranial sutures, can further complicate the clinical picture. Sagittal craniosynostosis, or scaphocephaly, results in an elongated head shape due to fusion along the sagittal suture (Vinchon *et al*., 2024). This condition often coexists with other neurodevelopmental anomalies, underscoring the importance of thorough imaging and physical examination in neonates with abnormal cranial morphology (Barkovich *et al*., 2012; Guerrini & Dobyns, 2014).

In resource-limited settings, like Ghana, early diagnosis and management of complex neonatal conditions such as jaundice, craniosynostosis, and lissencephaly remain challenging. Advanced imaging techniques, including MRI, are critical in such cases for identifying the underlying structural abnormalities, guiding clinical decisions, and improving outcomes (Shroff *et al*., 2010; Apolot *et al*., 2022). This case report describes a female neonate admitted to the Neonatal Intensive Care Unit (NICU) at the Komfo Anokye Teaching Hospital in Kumasi, Ghana. She presented with severe neonatal jaundice and cranial abnormalities consistent with sagittal craniosynostosis. MRI revealed features consistent with lissencephaly, emphasizing the role of neuroimaging in diagnosing and managing complex neonatal presentations (Gowda *et al*., 2021; Vanbuggenhout *et al*., 2022).

CASE REPORT

A female neonate less than one month old was admitted to the Neonatal Intensive Care Unit (NICU) at the Komfo Anokye Teaching Hospital in Kumasi, Ghana, presenting with visible jaundice. Laboratory investigations revealed significantly elevated bilirubin levels, indicating severe neonatal jaundice. Immediate phototherapy was initiated to lower bilirubin concentrations and reduce the risk of kernicterus. On physical examination, the neonate exhibited an abnormal cranial shape consistent with sagittal craniosynostosis. This was characterized by an elongated head (scaphocephaly) and palpable ridging along the sagittal suture, suggesting premature fusion of the sagittal cranial bones. These findings highlighted the need for additional imaging to assess for underlying structural abnormalities.

Investigation and Imaging Findings

The case of a female neonate admitted to the Neonatal Intensive Care Unit (NICU) at Komfo Anokye Teaching Hospital (KATH), Kumasi, Ghana, began with clinical findings of severe neonatal jaundice and abnormal cranial morphology. Laboratory investigations revealed markedly elevated bilirubin levels, necessitating prompt initiation of phototherapy to reduce bilirubin concentrations and mitigate the risk of kernicterus, a potentially irreversible neurological complication (Maisels & McDonagh, 2020). Physical examination further revealed an elongated cranial shape (scaphocephaly) and palpable ridging along the sagittal suture, features highly suggestive of sagittal craniosynostosis resulting from premature fusion of the sagittal cranial bones (Vinchon *et al*., 2018). Given these findings and concerns about underlying neurological anomalies, advanced imaging was deemed essential for further evaluation.

Initial ultrasound imaging suggested lobar holoprosencephaly (fig 2.0; A), a developmental forebrain malformation caused by incomplete division of the prosencephalon during early embryogenesis (Guerrini & Dobyns, 2014). However, due to the limitations of ultrasound in detecting subtle or complex brain malformations, magnetic resonance imaging (MRI) was performed to obtain more detailed anatomical information. MRI conducted on a Toshiba Achieva 1.5 Tesla scanner provided critical insights, revealing bilateral frontoparietal and temporal lissencephaly (fig 2.0; B), a condition characterized by a smooth cortical surface with absent gyration and sulcation. Lissencephaly results from impaired neuronal migration during brain development, often linked to genetic disorders such as tubulinopathies (Barkovich *et al*., 2015; Di Donato *et al*., 2017).

Further MRI findings included an absent septum pellucidum, a structure that separates the lateral ventricles and whose absence is frequently associated with midline brain anomalies like septo-optic dysplasia. Additionally, there was fusion of the basal ganglia with poor definition of the internal capsule (fig 3.0), a hallmark of disrupted neuronal organization often seen in cortical malformations. Notably, the MRI also demonstrated significant dilation of the lateral and third ventricles, consistent with ventriculomegaly, which likely resulted from impaired cerebrospinal fluid (CSF) dynamics due to the underlying brain malformations (Barkovich *et al*., 2015).

While the initial ultrasound findings suggested lobar holoprosencephaly, MRI provided greater diagnostic precision, confirming the presence of lissencephaly and associated abnormalities. The co-occurrence of neonatal jaundice and lissencephaly appeared coincidental, as lissencephaly is a congenital cortical malformation unrelated to bilirubin neurotoxicity (Maisels & McDonagh, 2020). However, the combination of sagittal craniosynostosis and ventricular dilation raised concerns regarding increased intracranial pressure, a potential complication requiring close neurosurgical monitoring.

MRI played a pivotal role in confirming the diagnosis and delineating the extent of the structural brain anomalies, enabling a more comprehensive clinical assessment and management plan. Early identification of these conditions facilitated targeted interventions, including phototherapy for hyperbilirubinemia and ongoing neurosurgical and neurological evaluations to

address craniosynostosis and developmental concerns. This case highlights the indispensable role of MRI in diagnosing complex congenital brain disorders and underscores the importance of multidisciplinary care in optimizing outcomes for affected neonates, particularly in resource-limited settings.

MRI Images and Findings

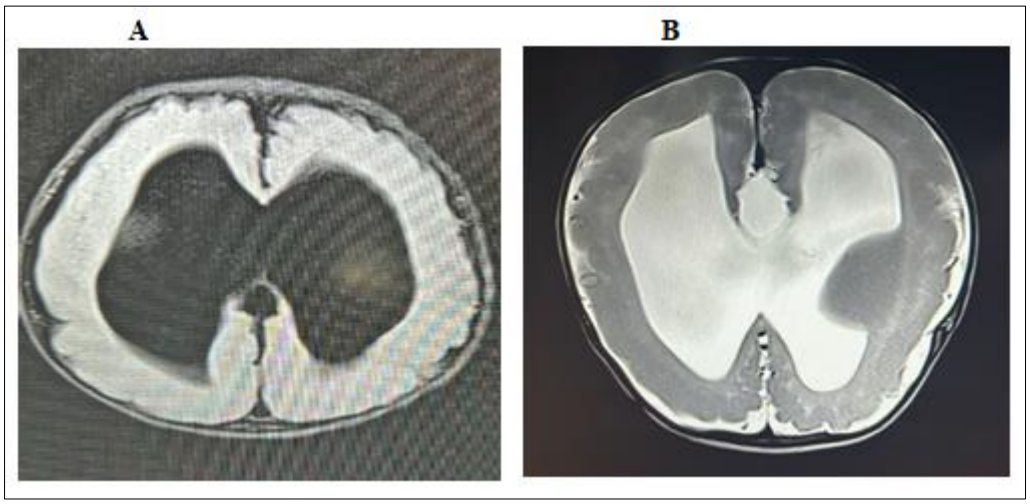

Figure 1.0: A & B: Axial FLAIR and a T2-weighted axial MRI images showing dilation of the lateral ventricles and fused basal ganglia with poor definition of internal capsule

Ultrasound vs MRI

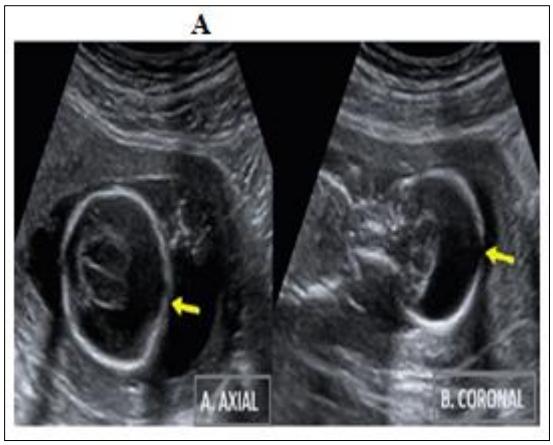

Figure 2.0: A: Axial and coronal ultrasound images showing findings suggestive of lobar holoprosencephaly

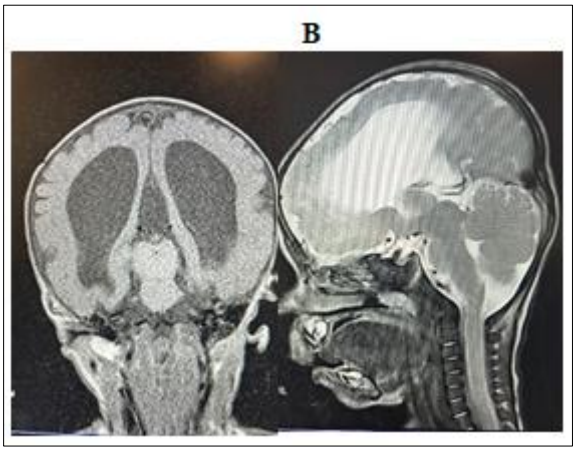

Figure 2.0: B: T1-weighted MRI coronal image showing bilateral frontoparietal and temporal lissencephaly and T2-weighted MRI sagittal image showing absence of septum pellucidum respectively

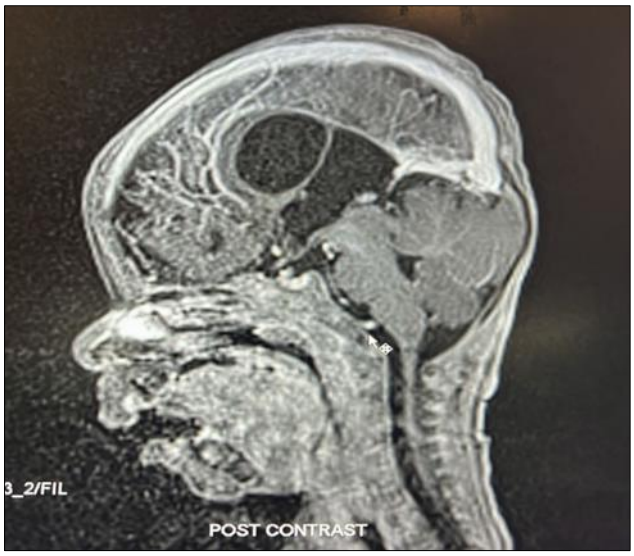

Figure 3.0: T1-weighted post-contrast MPRAGE showing smooth cortical surfaces consistent with lissencephaly, an absent septum pellucidum, and poorly defined basal ganglia with fused structures. Contrast enhancement highlights structural abnormalities and vascular details

Follow-up and Treatment Outcomes

Following the diagnosis of lissencephaly, sagittal craniosynostosis, and neonatal jaundice, a multidisciplinary treatment plan was initiated to address the immediate and long-term needs of the neonate. Immediate phototherapy was effective in reducing the elevated bilirubin levels, thereby lowering the risk of kernicterus and bilirubin-induced neurological damage. Serial bilirubin measurements were conducted during the course of phototherapy, and the neonate's levels showed a steady decline to within the normal range. This intervention successfully resolved the acute risk posed by neonatal jaundice, with no further signs of bilirubin encephalopathy observed during the hospitalization period.

In parallel, the abnormal cranial shape caused by sagittal craniosynostosis prompted close neurosurgical evaluation. While no immediate surgical intervention was deemed necessary, the neonate was placed on regular follow-up to monitor cranial growth and assess for signs of increased intracranial pressure. Neurosurgical assessments included periodic cranial imaging and head circumference measurements to detect any progression of craniosynostosis or complications such as hydrocephalus. If evidence of increased pressure or developmental delays were to emerge, cranial vault remodeling surgery would be considered as a corrective option.

Neurological follow-up was also initiated to monitor developmental milestones, given the presence of bilateral frontoparietal and temporal lissencephaly. Lissencephaly, a severe cortical malformation, predisposes affected individuals to a range of developmental issues, including motor delays, intellectual disabilities, and epilepsy. Regular neurological evaluations were scheduled to assess the neonate's motor skills, cognitive development, and overall growth. Parents and caregivers were educated on recognizing early signs of seizures or developmental delays, ensuring timely intervention when needed.

Follow-up imaging, including MRI scans, was planned at 6-month intervals to monitor brain development, ventricular size, and any progression of associated complications. These follow-up scans were critical in evaluating whether the ventriculomegaly identified during the initial imaging was stable or worsening, as increasing ventricular dilation could necessitate additional interventions, such as ventriculoperitoneal shunting.

At subsequent follow-up visits, the neonate demonstrated stable vital signs and no acute complications. Although delays in motor milestones were anticipated due to the lissencephaly, early intervention strategies, such as physical and occupational therapy, were recommended to optimize developmental outcomes. Supportive care, including family counseling and access to rehabilitation services, was emphasized to enhance the quality of life and address the long-term needs of the child.

To conclude, the combination of effective management of neonatal jaundice, close neurosurgical monitoring for craniosynostosis, and ongoing neurological care allowed for comprehensive treatment of this complex case. While lissencephaly remains a lifelong condition requiring ongoing care, the follow-up plan aims to ensure timely interventions, monitor neurological status, and provide developmental support for the neonate's growth and well-being.

DISCUSSIONS

This case highlights the complex interplay of multiple neurological and systemic conditions, including lissencephaly, sagittal craniosynostosis, and neonatal jaundice, which were diagnosed in a female neonate admitted to the Neonatal Intensive Care Unit (NICU) at Komfo Anokye Teaching Hospital (KATH), Kumasi, Ghana. The coexistence of these conditions presents unique diagnostic and management challenges, particularly in resource-limited settings. Comprehensive investigations, including laboratory tests and advanced neuroimaging with magnetic resonance imaging (MRI), played a crucial role in providing an accurate diagnosis and guiding clinical management.

Lissencephaly, identified through MRI, is a severe cortical malformation characterized by a lack of normal brain gyration and sulcation, resulting from defective neuronal migration during embryogenesis (Barkovich *et al*., 2012; Di Donato *et al*., 2017). The presence of bilateral frontoparietal and temporal lissencephaly, along with an absent septum pellucidum and fused basal ganglia with poor internal capsule definition, aligns with the findings reported in cases of tubulinopathies, a genetic disorder affecting microtubule function (Guerrini & Dobyns, 2014). Such malformations frequently lead to developmental delays, intellectual disabilities, and epilepsy, as described by Barkovich *et al*., (2012) and Russ *et al*., (2021). The ventriculomegaly observed, with dilation of the lateral and third ventricles, is likely secondary to the disrupted cerebrospinal fluid (CSF) pathways caused by abnormal brain structure.

The coexistence of sagittal craniosynostosis further complicates this case. Craniosynostosis, specifically sagittal craniosynostosis or scaphocephaly, results from the premature fusion of the sagittal suture, leading to elongated head morphology and increased risk of raised intracranial pressure (Vinchon *et al*., 2024). This condition can occur in isolation or as part of a syndrome, making imaging critical for differentiating between primary and secondary causes of cranial abnormalities. In this neonate, MRI findings confirmed the abnormal cranial shape and excluded other associated midline brain malformations, apart from the lissencephaly. Early neurosurgical evaluations and planned follow-up imaging are essential to monitor for complications such as hydrocephalus and elevated intracranial pressure (Shroff *et al*., 2010).

The third condition, neonatal jaundice, posed an immediate risk of bilirubin neurotoxicity. Elevated bilirubin levels, as observed in this case, can cross the blood-brain barrier and cause kernicterus, a preventable but irreversible form of bilirubin encephalopathy (Shapiro, 2010; Maisels & McDonagh, 2008). Early identification and management of jaundice with phototherapy were critical in preventing acute bilirubin toxicity. Kernicterus, if untreated, can exacerbate neurological outcomes in neonates with pre-existing conditions like lissencephaly (Bhutani *et al*., 2004). However, in this case, neonatal jaundice and lissencephaly appeared coincidental, as no direct etiological link exists between the two conditions.

The presence of lobar holoprosencephaly on initial ultrasound further added complexity to the diagnostic process. Holoprosencephaly, a forebrain malformation resulting from failed cleavage of the prosencephalon, shares overlapping imaging and clinical features with lissencephaly, including midline brain abnormalities such as absence of the septum pellucidum (Hayward *et al*., 1991; Vanbuggenhout *et al*., 2022). Advanced MRI allowed for differentiation between these conditions, providing a definitive diagnosis of lissencephaly. This underscores the importance of MRI in evaluating neonates with suspected structural brain anomalies (Shroff *et al*., 2010; Hüppi & Barnes, 1997).

From a pathophysiological perspective, lissencephaly is often associated with mutations in genes responsible for neuronal migration, such as LIS1 and TUBA1A, which affect microtubule dynamics (Di Donato *et al*., 2017; Guerrini & Dobyns, 2014). The resultant malformations of cortical development predispose affected individuals to a broad spectrum of neurological sequelae, including epilepsy, motor deficits, and intellectual disability (Guerrini *et al*., 2014; Russ *et al*., 2021). These developmental complications necessitate early intervention strategies, such as physical therapy and ongoing neurological monitoring, as highlighted in this case.

The findings in this case align with previous reports on the diagnostic value of MRI in detecting congenital brain malformations. MRI remains the gold standard for evaluating cortical malformations and ventricular abnormalities, offering superior anatomical detail compared to ultrasound and CT (Barkovich *et al*., 2012; Averill *et al*., 2015). Additionally, the presence of ventriculomegaly necessitates longitudinal imaging follow-up to monitor for hydrocephalus, a common complication in lissencephaly (Shroff *et al*., 2010; Apolot *et al*., 2022).

This case underscores the significance of early imaging, particularly MRI, in diagnosing complex congenital neurological anomalies like lissencephaly and craniosynostosis. The timely management of neonatal jaundice through phototherapy mitigated immediate risks, while neurosurgical and neurological follow-up remains essential to monitor cranial development, prevent complications, and provide developmental support. Comprehensive multidisciplinary care is critical for optimizing outcomes in neonates with coexisting neurological and systemic conditions. This case also highlights the importance of advanced imaging modalities in resource-limited settings for improving diagnostic accuracy and guiding treatment strategies.

CONCLUSION

This case highlights the critical role of advanced imaging, particularly MRI, in the diagnosis and management of complex congenital brain anomalies. The detailed MRI findings were pivotal in the diagnosis of bilateral frontoparietal and temporal lissencephaly. These findings informed a comprehensive management plan, including neurological evaluations, early therapeutic interventions, hydrocephalus surveillance, and interdisciplinary follow-up care.

The successful application of MRI in this case shows the importance of early access to advanced diagnostic tools in resource-limited settings like Ghana, where early and accurate identification of such anomalies is often challenging. The case also emphasizes the necessity of a multidisciplinary approach, integrating neonatology, neurology, neurosurgery, and developmental care, to address the multifaceted needs of neonates with severe congenital conditions.

Ultimately, this case demonstrates how leveraging advanced imaging technology and collaborative care can optimize outcomes for patients with complex medical presentations, even in low-resource environments. It serves as a call to action for improving healthcare infrastructure and access to advanced diagnostic modalities to enhance care for vulnerable populations.

Patient Consent: Verbal informed consent for this publication of this case report was obtained from the parents of the patient.

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