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Case Report

Congenital external hydrocephalus: A rare presentation of lobar holoprosencephaly in a neonate ☆

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ABSTRACT

External hydrocephalus is a rare condition characterized by abnormal cerebrospinal fluid (CSF) accumulation in the subarachnoid space, often associated with developmental anomalies. Lobar holoprosencephaly, a milder form of holoprosencephaly, can manifest with hydrocephalus due to its impact on brain development and CSF dynamics. This case report describes a neonate with congenital external hydrocephalus secondary to lobar holoprosencephaly, highlighting the diagnostic imaging findings and management approach. A neonate presented with progressive macrocephaly, irritability, altered sensorium, and poor feeding. Antenatal ultrasound at 32 weeks of gestation revealed macrocephaly and hydrocephalus, leading to a cesarean delivery at 38 weeks. Physical examination showed an occipitofrontal circumference of 45 cm, exceeding the 97th percentile for age. Magnetic resonance imaging (MRI) revealed fused frontal horns of the lateral ventricles, hypoplasia of the posterior corpus callosum, and extensive extra-axial CSF accumulation compressing the brain parenchyma. The CSF collection showed complete suppression on FLAIR imaging, confirming its nature, and a cortical vein sign indicated an enlarged subarachnoid space rather than a subdural hygroma. A diagnosis of lobar holoprosencephaly with congenital external hydrocephalus was made. The patient underwent peritoneal shunting to alleviate intracranial pressure, significantly reducing head circumference to 38 cm. Postoperative recovery was uneventful, and the parents were counseled on genetic testing and long-term follow-up. This case underscores the importance of detailed neuroimaging in differentiating external hydrocephalus from other pathologies and highlights the role of surgical intervention in improving outcomes. Early diagnosis and a multidisciplinary approach are vital for managing complex congenital anomalies such as lobar holoprosencephaly.

☆ Competing Interests: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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Background

Hydrocephalus, a condition resulting from an imbalance between the production and absorption of cerebrospinal fluid (CSF), manifests with increased intracranial pressure, ventricular dilation, and, often, progressive macrocephaly [1]. While hydrocephalus can arise from a myriad of causes, congenital forms are predominantly associated with structural brain malformations, genetic syndromes, or intrauterine insults [2]. Among its less common variants is external hydrocephalus, characterized by abnormal CSF accumulation in the subarachnoid space, frequently leading to compression of brain parenchyma. This rare presentation is more often observed in neonates and is closely linked with developmental anomalies such as holoprosencephaly [3].

Holoprosencephaly is a complex congenital disorder that arises from incomplete cleavage and differentiation of the forebrain during early embryogenesis, typically between the third and fourth weeks of gestation. Its severity ranges from alobar, where there is complete fusion of the cerebral hemispheres, to lobar holoprosencephaly, where separation is incomplete but less severe [4]. Lobar holoprosencephaly is often associated with abnormalities in CSF pathways due to distorted anatomy, resulting in hydrocephalus. These anomalies disrupt CSF dynamics, causing either internal or external hydrocephalus depending on the site of obstruction or impaired absorption [5].

Neuroimaging plays a pivotal role in diagnosing external hydrocephalus, especially in distinguishing it from other conditions like subdural hygroma, which can present similarly [6]. MRI findings characteristic of external hydrocephalus include enlarged subarachnoid spaces with suppressed CSF signals on fluid-attenuated inversion recovery (FLAIR) imaging, cortical vein sign (indicating an enlarged subarachnoid space rather than a subdural collection), and associated structural abnormalities such as ventricular fusion or corpus callosum hypoplasia [7]. These imaging features are critical for timely diagnosis and differentiation from other conditions that require distinct management strategies.

Untreated hydrocephalus, regardless of its origin, can lead to irreversible brain damage, developmental delays, and long-term neurological sequelae. In cases of external hydrocephalus, ventriculoperitoneal (VP) shunting remains the mainstay of treatment, effectively reducing intracranial pressure and alleviating symptoms [8]. Surgical intervention is often complemented by genetic evaluation and counseling, particularly for cases associated with structural anomalies such as holoprosencephaly, which is linked with chromosomal abnormalities (e.g., trisomy 13) or mutations in SHH, ZIC2, and SIX3 genes [9].

This case report aims to underscore the diagnostic, therapeutic, and prognostic challenges associated with external hydrocephalus, particularly in lobar holoprosencephaly. By providing detailed insights into the clinical and imaging features, this report contributes to the existing literature. It highlights the importance of a multidisciplinary approach in managing such complex congenital anomalies.

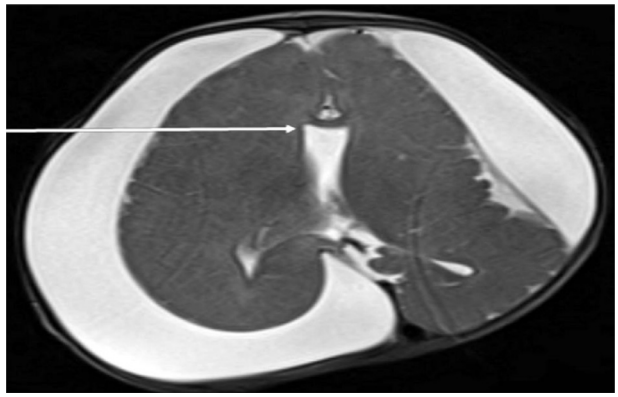

Fig 1 – Axial T2-weighted image depicting fused and rudimentary frontal horns of the bilateral lateral ventricles (indicated by white arrow) with a wide communication between the fused segment and the third ventricle.

Case presentation

A neonate was brought to the hospital by her parents with concerns of a progressively enlarging head size, irritability, altered sensorium, and poor feeding. The infant was born via cesarean section at 38 weeks of gestation due to prenatal detection of macrocephaly and potential fetal distress. The antenatal history was significant, with an ultrasound performed at 32 weeks of gestation revealing macrocephaly and hydrocephalus. The fetal head circumference measured 33.5 cm, exceeding the 97th percentile for the gestational age. These findings led to a planned cesarean delivery to mitigate complications during labor.

Upon physical examination at presentation, the infant displayed an occipitofrontal circumference of 45 cm, further confirming macrocephaly, as it was significantly above the 97th percentile for age. Additionally, the neonate exhibited clinical features including frontal bossing, a downward gaze (setting-sun sign), and drowsiness, all indicative of raised intracranial pressure. These findings warranted an immediate neuroimaging evaluation to determine the underlying cause.

Magnetic resonance imaging (MRI) was performed to elucidate the etiology of the hydrocephalus. The imaging revealed fused and rudimentary frontal horns of the bilateral lateral ventricles, with an abnormally wide communication between the fused segment and the third ventricle Fig. 1. The sagittal T2-weighted images showed nonvisualization and hypoplasia of the posterior aspect of the corpus callosum and the posterior portion of its body Fig. 2. Further imaging identified an extensive extra-axial cerebrospinal fluid (CSF) collection along the bilateral fronto-parieto-occipital and temporal regions, exerting significant compression on the brain parenchyma Fig. 3.

The fluid collection demonstrated complete suppression on fluid-attenuated inversion recovery (FLAIR) imaging Fig. 4, confirming the diagnosis of cerebrospinal fluid rather than another type of fluid, such as a subdural hygroma. Additionally, a vessel traversing the enlarged subarachnoid space (cortical vein sign) further supported the finding of an enlarged subarachnoid space rather than a subdural pathology. The in-

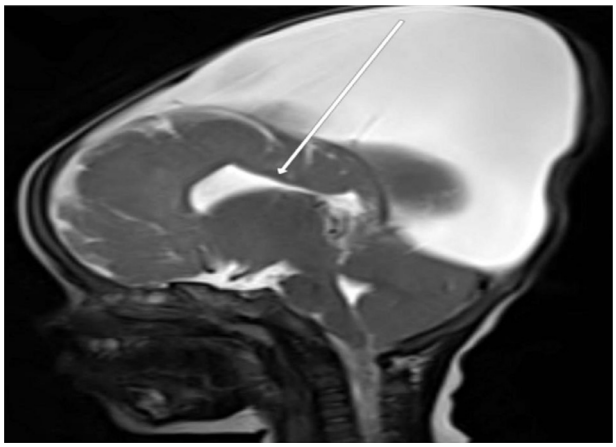

Fig 2 – Sagittal T2-weighted image demonstrating the absence or hypoplasia of the posterior aspect of the corpus callosum (indicated by the white arrow).

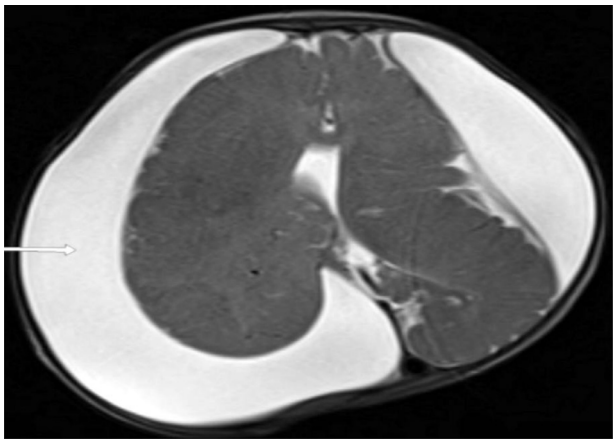

Fig 3 – Axial T2-weighted image illustrating extra-axial cerebrospinal fluid (CSF) intensity collection (white arrow) in the bilateral fronto-parieto-occipital and temporal regions.

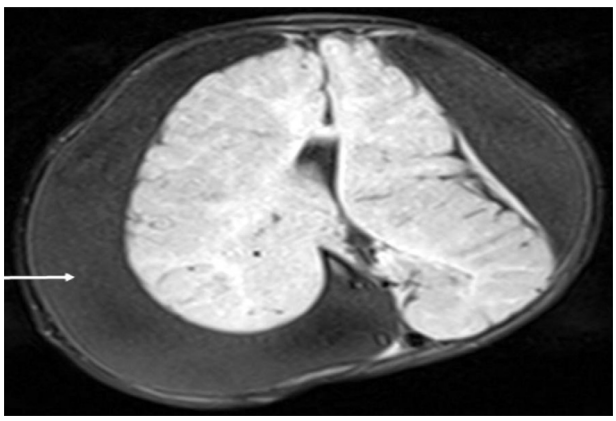

Fig 4 – Axial FLAIR (Fluid Attenuated Inversion Recovery) image showing complete suppression of fluid collection (indicated by the white arrow), confirming it as cerebrospinal fluid (CSF).

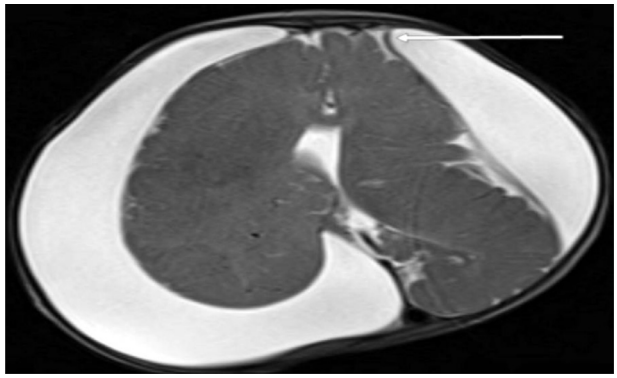

Fig 5 – Axial T2-weighted image showing a vessel (indicated by the white arrow) traversing the enlarged subarachnoid space, demonstrating the cortical vein sign and confirming the presence of an enlarged subarachnoid space.

terhemispheric fissure and falx cerebri were well visualized, helping rule out other differential diagnoses Fig. 5.

Based on these findings, the infant was diagnosed with lobar holoprosencephaly associated with congenital external hydrocephalus. Lobar holoprosencephaly is a developmental anomaly characterized by incomplete separation of the cerebral hemispheres, contributing to the abnormal CSF accumulation in this case.

A referral to neurosurgery was made, and the patient underwent a peritoneal shunt placement to relieve the external hydrocephalus. Postoperative measurements revealed a reduction in the occipitofrontal circumference to 38 cm, reflecting successful fluid drainage and decreased intracranial pressure. The infant demonstrated clinical improvement following the procedure, with better sensorium and feeding patterns.

The parents were counseled extensively about the diagnosis and its implications. They were informed about the need for long-term monitoring, potential complications, and regular follow-up with a multidisciplinary team, including neurology and genetics specialists. Genetic counseling and testing were recommended to investigate potential underlying syndromic causes, given the association of holoprosencephaly with genetic anomalies.

This detailed case highlights the importance of early detection, accurate radiological interpretation, and prompt surgical management in achieving optimal outcomes for complex congenital conditions such as external hydrocephalus with lobar holoprosencephaly.

Discussion

External hydrocephalus, a rare condition in neonates, is characterized by an abnormal accumulation of cerebrospinal fluid (CSF) in the subarachnoid space, often leading to symptoms such as macrocephaly, irritability, and poor feeding [10]. The condition can result from various etiologies, including de-

velopmental anomalies like holoprosencephaly. Holoprosencephaly represents a spectrum of forebrain malformations caused by incomplete midline division during embryogenesis, with lobar holoprosencephaly being its mildest form [11]. In this case, external hydrocephalus was linked to lobar holoprosencephaly, highlighting the intricate interplay between developmental brain anomalies and CSF dynamics.

Role of imaging in diagnosis

Neuroimaging, particularly magnetic resonance imaging (MRI), was pivotal in diagnosing this case. The MRI findings demonstrated fused frontal horns of the lateral ventricles, a wide communication with the third ventricle, hypoplasia of the posterior corpus callosum, and a significant extra-axial CSF collection compressing the brain parenchyma [12]. The fluid's complete suppression on fluid-attenuated inversion recovery (FLAIR) imaging confirmed its CSF nature. Additionally, the cortical vein sign—a vessel traversing the enlarged subarachnoid space—was crucial in distinguishing the condition from a subdural hygroma. Accurate imaging interpretation is essential for differentiating external hydrocephalus from other conditions, as misdiagnosis can lead to inappropriate management [13]. Studies have shown that advanced imaging techniques, including diffusion-weighted imaging and volumetric analysis, can provide deeper insights into brain structure and CSF dynamics in congenital conditions like holoprosencephaly. These modalities help assess structural anomalies' severity, guiding prognosis and management [14].

Management and surgical intervention

Management strategies for external hydrocephalus vary depending on the severity of symptoms and the underlying cause. Peritoneal shunting was performed in this patient, successfully reducing intracranial pressure and head circumference. Shunting remains the cornerstone of treatment for symptomatic hydrocephalus, as it effectively mitigates intracranial pressure and prevents progressive neurological deterioration. A reduction in head circumference postshunting, as observed in this case, correlates with improved clinical outcomes [15]. However, surgical management is not without risks. Complications such as infection, shunt malfunction, or over-drainage can occur, necessitating close postoperative monitoring. Studies have reported success rates of 80%-90% for peritoneal shunting in neonates with hydrocephalus, with long-term outcomes largely dependent on the severity of the associated anomalies [16].

Genetic considerations and counseling

Holoprosencephaly is frequently associated with genetic abnormalities, including chromosomal aneuploidies (e.g., trisomy 13) and mutations in key developmental genes such as SHH, ZIC2, and SIX3. These genetic factors disrupt normal forebrain development, resulting in the spectrum of malformations observed in holoprosencephaly. In this case, genetic counseling and testing were recommended to identify potential hereditary factors and assess recurrence risks in future pregnancies [4]. Genetic testing also aids in providing a clearer prognosis. For example, SHH mutations are associated with a variable phenotype, while anomalies linked to trisomy 13 typically have a poor prognosis due to multisystem involvement. Understanding the genetic underpinnings of holoprosencephaly enables personalized care and informed decision-making for families [17].

Prognosis and long-term outcomes

The prognosis for patients with external hydrocephalus depends on the severity of associated brain anomalies. Lobar holoprosencephaly, being a milder form, generally allows for better outcomes compared to more severe variants like alobar holoprosencephaly. However, persistent developmental delays, motor deficits, and cognitive challenges remain concerns. Regular follow-ups with a multidisciplinary team, including neurology, developmental pediatrics, and genetics, are crucial for addressing these issues [18]. This case underscores the importance of early diagnosis and timely intervention in managing complex congenital anomalies. A multidisciplinary approach combining radiological, surgical, and genetic expertise significantly improves outcomes. By documenting this rare association, we aim to contribute to the growing understanding of external hydrocephalus and its management in neonates with holoprosencephaly.

Conclusion

This case highlights the rare presentation of congenital external hydrocephalus associated with lobar holoprosencephaly in a neonate, emphasizing the importance of early diagnosis and intervention. Detailed neuroimaging was pivotal in accurately identifying the underlying etiology, differentiating external hydrocephalus from other potential causes such as subdural hygroma. The findings, including fused frontal horns, hypoplasia of the corpus callosum, and cortical vein sign, underscored the developmental abnormalities contributing to the condition. Prompt surgical management through peritoneal shunting effectively alleviated intracranial pressure, reducing the occipitofrontal circumference and improving the patient's clinical status. This case underscores the need for a multidisciplinary approach, combining radiological, neurosurgical, and genetic expertise, to optimize outcomes for neonates with complex congenital anomalies. Comprehensive parental counseling and long-term follow-up are essential components of care, addressing potential complications and supporting developmental needs. By documenting this unique case, we aim to enhance understanding and awareness of external hydrocephalus as a rare manifestation of lobar holoprosencephaly, contributing to improved recognition and management of similar presentations in clinical practice.

Patient consent

Written informed consent was obtained from the patient for the publication of this case report.

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