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Radiological Imaging in Hydranencephaly: A Case Report

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Abstract

Hydranencephaly is a rare congenital anomaly characterized by the near-complete absence of the cerebral hemispheres with preservation of the brainstem and basal ganglia. Radiological imaging plays a crucial role in the antenatal and postnatal diagnosis, management, and prognostication of this condition. This abstract provides a comprehensive review of the radiological findings in hydranencephaly.

Prenatal ultrasound typically reveals a lack of cerebral tissue with the presence of fluid-filled cranial vaults. Magnetic resonance imaging (MRI) further delineates the extent of brain tissue loss, often showing remnants of the basal ganglia and thalami. Postnatally, computed tomography (CT) scans confirm the absence of cerebral hemispheres and may reveal calcifications in the basal ganglia.

Radiological findings aid in distinguishing hydranencephaly from other brain malformations such as anencephaly, holoprosencephaly, and porencephaly. Additionally, they assist in counselling families regarding prognosis and guiding clinical management decisions.

This abstract highlights the importance of radiological imaging in the diagnosis and management of hydranencephaly, emphasizing its role in facilitating early intervention and supportive care for affected individuals.

Keywords: Angiography, Hydranencephaly, Sigmoid Sinuses, Visualization

Case report

A 7-month-old female infant presented with macrocephaly and developmental delay since birth. Past history of Post-natal neuro-sonogram revealed hydrocephalus and suggested further radiological evaluation but the patient underwent homeopathic treatment. Now patient underwent a neuro-sonogram which revealed severely dilated ventricles and a fluid-filled cranial vault. There is minimal visualization of the brain parenchyma secondary to the large volume of fluid. Further patient underwent an MRI scan showing, Bilateral supratentorial parenchyma replaced by CSF fluid with small residual brain parenchyma in the bilateral temporo-occipital regions. There is a mass effect over the posterior fossa structures. However posterior fossa structures appear normal. Midline falx is seen.

Bilateral basal ganglia appear separated. 4th ventricle appears normal.

MR angiography reveals bilateral normal flow in the extracranial and intracranial portions of the internal carotid artery. V4 segment of bilateral vertebral and basilar artery shows normal flow. Absent flow was noted in the bilateral anterior, middle, and posterior cerebral arteries.

MR venography reveals Hypoplastic bilateral transverse and sigmoid sinuses. Superior and inferior sagittal sinuses show absent flow.

Figure 1: Clinical picture of 7 months old infant presented with macrocephaly

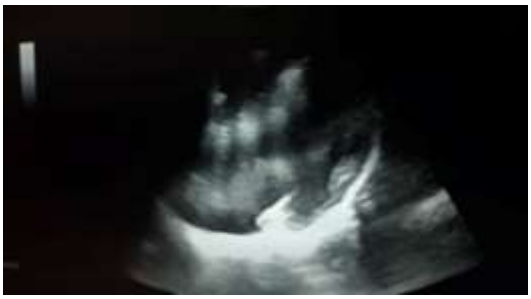

Figure 2: Neuro-sonogram shows large volume of fluid-filled cranial vault with minimal visualization of the brain parenchyma

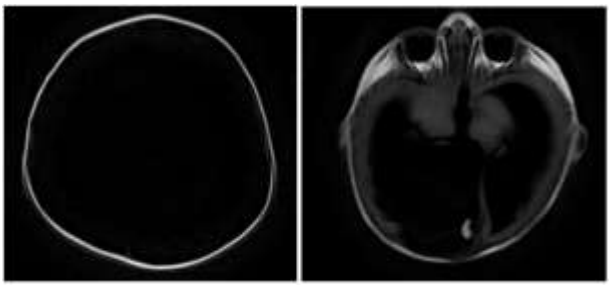

Figure 3 and 4: T1W MRI axial section shows supra tentorial cerebral parenchyma replaced by CSF density fluidwith small residual brain parenchyma in the bilateral temporo-occipital regions. Bilateral basal ganglia appear normal and separated

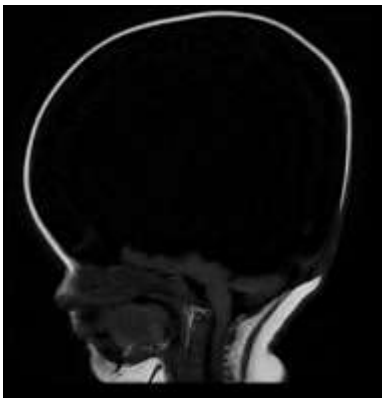

Figure 5: T1W MRI sagittal section showssupra tentorial cerebral parenchyma replaced by CSF density fluid. However, posterior fossa and brain stem structures appear normal.

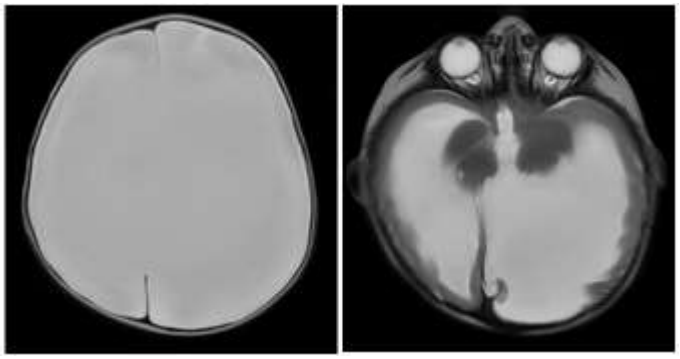

Figure 6 and 7: T2W MRI axial section shows presence of mid line falx.

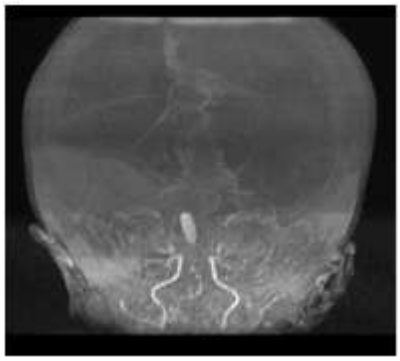

Figure 8: MR angiogram showing bilateral normal flow in an extracranial and intracranial portion of the internal carotid artery with the absent flow in the bilateral anterior, middle, and posterior cerebral arteries.

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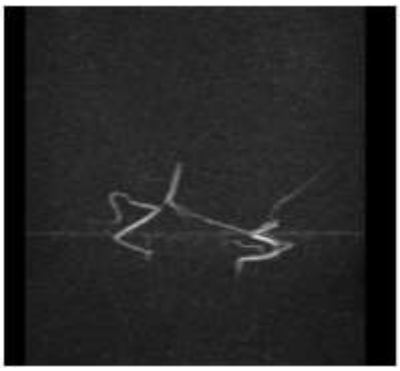

Figure 9: MR venogram showing hypoplastic bilateral transverse and sigmoid sinuses with absent flow in superior and inferior sagittal sinuses.

Discussion

Hydranencephaly (HE) represents a rare prenatal condition characterized by the near-complete absence of the cerebral hemispheres, replaced by membranous sacs filled with cerebrospinal fluid, glial tissue, and ependyma. Typically, structures such as the cerebellum, midbrain, basal ganglia, thalami, and choroid plexus remain unaffected. Hemihydranencephaly, though less common, presents with the unilateral absence of one cerebral hemisphere.Various etiologies have been proposed, with bilateral occlusion of the supraclinoid segment of the internal carotid arteries being the predominant hypothesis, supported by angiographic evidence of aplastic or hypoplastic arteries. Other theories suggest in utero destruction of formed brain structures, potentially due to encephaloclastic mechanisms, or dysontogenetic processes disrupting organogenesis. Molecular dysfunctions, including mutations such as COL4A1 or PI3K-Akt3-mTOR mutations, are also being explored.

Numerous predisposing factors have been implicated, including intrauterine infections (e.g., toxoplasmosis), viral infections (e.g., Zika, rubella, cytomegalovirus), certain medications (e.g., warfarin, sodium valproate), maternal irradiation, toxic exposures (e.g., cocaine, smoking), and various genetic abnormalities. Differential diagnosis poses challenges, with conditions such as alobar holoprosencephaly, severe hydrocephalus, and porencephalic cysts being considered. Alobar holoprosencephaly typically presents with midline malformations, absence of a falx, and a distinct pancake-shaped residual cortex. In severe hydrocephalus, a cortical rim remains intact, and the middle cerebral arteries are preserved, while porencephalic cysts are often associated with ischemic infarcts in the middle cerebral artery territory, leading to localized cortical destruction. Diagnostic modalities for HE include ultrasound, CT, and MRI, with MRI and magnetic resonance angiography (MRA) being the preferred diagnostic tools.

Prognosis for individuals with HE is generally poor, with most affected patients succumbing in utero or within the first year of life. Prolonged survival is exceedingly rare.

Conclusion

Hydranencephaly represents a congenital neurodevelopmental abnormality characterized by the absence of the cerebral hemispheres while infratentorial structures remain intact. This condition is typically associated with bilateral occlusion of the internal carotid arteries (ICAs). Diagnosis can occur either during prenatal or postnatal stages through ultrasound, CT scans, or MRI. Radiological findings often reveal rudimentary brain structures, variable presence of the falx cerebri, and complete or near-complete absence of the cerebral hemispheres. Regrettably, there is currently no known cure for hydranencephaly, and the prognosis for affected individuals tends to be unfavorable.

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