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Case Report

Radiology Section

CT Unveils a Striking Case of Hydranencephaly: A Case Report

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ABSTRACT

A rare congenital brain condition known as hydranencephaly is characterised by the near-total loss of the cerebral hemispheres, which are replaced by cerebrospinal fluid, although the brainstem and cerebellum are left intact. It is usually diagnosed in infancy; however, uncommon presentations can also occur. We describe a child who experienced frequent Generalised Tonic-Clonic Seizures (GTCS), drowsiness, and dyspnoea across four days, as well as a history of steady weight loss since birth. Clinical examination revealed an underlying neurological condition, and non-contrast Computed Tomography (CT) of the brain showed almost complete bilateral loss of cerebral parenchyma, except for the retained falx cerebri, thalami, brainstem, and cerebellum, confirming the diagnosis of hydranencephaly. This case emphasises the necessity of looking for hydranencephaly in neonates with unexplained seizures and developmental failure, as well as the critical role of radiological imaging, particularly CT and Magnetic Resonance Imaging (MRI), in attaining timely diagnosis, parental counselling, and appropriate care.

Keywords: Cerebral agenesis, Computed tomography Congenital brain anomaly, Paediatric neuroimaging, Seizures

CASE REPORT

A two-month-old male newborn presented to the emergency room with symptoms of repeated bouts of GTCS for four days. The convulsions were accompanied by postictal sleepiness and sporadic episodes of dyspnoea. The parents also claimed a gradual weight loss from birth. There was no evidence of fever, trauma, or recent infections.

Perinatal history revealed that the infant was born at term via normal vaginal birth, with no prenatal or intranatal complications. Details regarding antenatal ultrasonography were not available; hence, it could not be ascertained whether the anomaly was detected prenatally. The infant's birth weight was not documented in the available records; however, he appeared undernourished at presentation, with a weight well below the third percentile for age. The child had been less active since birth, with poor feeding and irritability. The parents were not consanguineous, and there was no family history of congenital malformations, neurological problems, or other conditions. The infant had an APGAR score of 8 and 9 at one and five minutes, respectively. Cardiovascular, respiratory, abdominal, and genitourinary examinations were within normal limits.

Clinical examination revealed that the infant was undernourished and seemed lethargic, with a weight that was well below the third percentile for age. A head circumference measurement revealed macrocephaly, along with a complete and tight anterior fontanelle [Table/Fig-1]. Reduced deep tendon reflexes, decreased spontaneous limb movements, and generalised hypotonia were found during the neurological test. All of the pupils responded to the light equally. There were no signs of craniofacial dysmorphism.

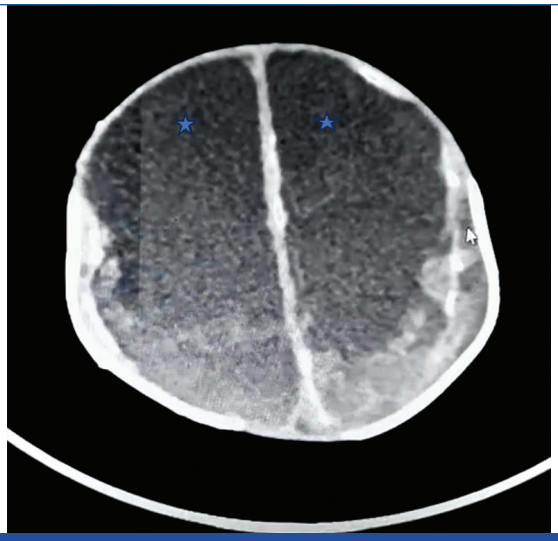
Initial laboratory investigations evaluated possible metabolic or infectious causes of seizures. The following findings were noted:

- Complete Blood Count (CBC): Within normal limits
- Serum electrolytes (Na+, K+, Ca2+): Normal
- Random blood glucose: 88 mg/dL
- C-Reactive Protein (CRP) and procalcitonin: Not elevated
- Liver and renal function tests: Within normal limits
- Serum lactate and ammonia: Normal
- Blood and urine cultures: Negative

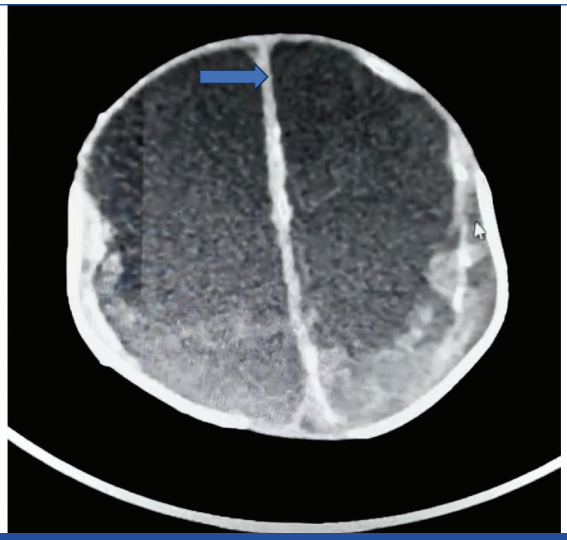
[Table/Fig-1]: Clinical image of the patient showing macrocephaly.

- Toxoplasmosis, Rubella, Cytomegalovirus (CMV), Herpes simplex (TORCH) screening: Non-reactive
- Arterial Blood Gas (ABG): Mild metabolic acidosis with respiratory compensation

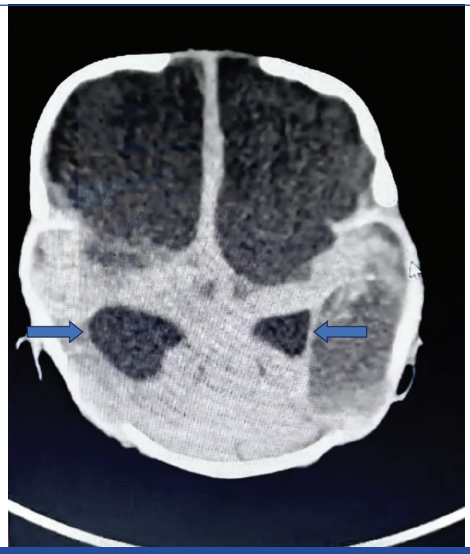
Given persistent seizures and abnormal neurological findings, a non-contrast CT scan of the brain was performed. CT revealed that the bilateral cerebral parenchyma is replaced by a Cerebrospinal Fluid (CSF) density space [Table/Fig-2], with an intact interhemispheric fissure [Table/Fig-3]. There is a normal formation of the ventricular system, with mild dilatation of the ventricular system (dilated occipital horns of bilateral lateral ventricles) [Table/Fig-4].



[Table/Fig-2]: CT brain axial section showing CSF fluid density space replacing bilateral cerebral parenchyma (blue star).



[Table/Fig-3]: CT brain axial section showing intact interhemispheric fissure (blue arrow).



[Table/Fig-4]: CT brain axial section showing dilated occipital horns of bilateral lateral ventricles (blue arrows).

A diagnosis of hydranencephaly was established based on clinical and radiological correlation. The patient was managed with intravenous phenobarbital (loading dose of 20 mg/kg, followed by a maintenance dose of 3-4 mg/kg/day) for five days, along with supportive oxygen therapy and nutritional rehabilitation. A multidisciplinary referral was made to paediatric neurology, neurosurgery, and clinical genetics for further evaluation and supportive care planning. The prognosis and long-term care implications were discussed with the parents, with emphasis on the poor neurodevelopmental outcomes and need for supportive therapy. Genetic counselling was recommended to investigate any hereditary or chromosomal correlations.

DISCUSSION

Hydranencephaly is an encephaloclastic disorder in which the cerebral hemispheres are replaced with cerebrospinal fluid and necrotic debris, which is then covered by leptomeninges. The cerebral cortex is usually missing; however, a portion of the occipital lobe may be maintained [1]. The midbrain, thalamus, basal ganglia, choroidal plexus, cerebellum, and brainstem are often enclosed within the skull.

Hydranencephaly is exceedingly rare, with an estimated incidence of approximately 1 in 10,000 live births and accounts for less than 1% of all intracranial congenital malformations [2].

Hydranencephaly's aetiology is complex, and numerous theories have been proposed to explain its occurrence. The most prevalent aetiology identified is the occlusion of the supraclinoid portion of the bilateral internal carotid arteries, resulting in ischaemic degeneration of structures fed by them [3]. Myers RE investigated the aetiology of hydranencephaly using laboratory monkeys. In his work, monkey foetuses at various gestational ages had their bilateral carotid arteries and jugular veins in the neck ligated. These foetuses were then returned to the uterus, carried to term, and delivered. Examination of the young monkey brains demonstrated hydranencephaly, resulting from a vascular shutdown, particularly during early gestational age [4]. Hydranencephaly may occasionally be associated with conditions such as Fowler syndrome, vascular malformations, or congenital infections, including TORCH group pathogens [2]. However, no such associations were found in the present case.

Literature implies the closure of the internal carotid arteries due to a transient spasm rather than direct occlusion, resulting in ischaemic damage of specific brain regions [5]. Other aetiologies of hydranencephaly include intrauterine infections that lead to the local destruction of brain tissue, such as congenital toxoplasmosis, or other viral infections (adenovirus, cytomegalovirus, enterovirus, Epstein-Barr virus, herpes simplex virus, parvovirus, and respiratory syncytial viruses) [6]. Another aetiology that has emerged is maternal exposure to carbon monoxide or butane gas, which can result in foetal hypoxia, leading to extensive tissue necrosis with cavitations, resorption of necrotised tissue, and necrotising vasculitis [7].

The idea of Internal Carotid Artery (ICA) occlusion is the most frequently accepted explanation for hydranencephaly, as an intact blood supply is required for normal foetal brain development. Early ICA occlusion can cause blockage of the downstream arteries that supply the cerebral hemispheres, resulting in a brain infarction and subsequent reabsorption of the wounded brain tissue as cerebrospinal fluid replaces the infarcted brain [6].

Hydranencephaly can be detected with any imaging technique that examines the foetal or newborn brain. Imaging findings include macrocephaly in utero, the lack of the supratentorial brain, and a massive cerebrospinal fluid collection in the cranial vault. The presence of the third ventricle varies. The falx cerebri may be partial, but it is usually present and can assist in distinguishing between hydranencephaly and alobar holoprosencephaly [8].

Severe hydrocephalus, alobar holoprosencephaly, and porencephaly were the primary differential diagnoses considered in this instance. These conditions can all manifest with seizures, developmental delay, and macrocephaly. Large ventricular dilatation may result from severe hydrocephalus; however, in contrast to hydranencephaly, which retains the falx cerebri and a thin rim of compressed cerebral cortex, severe hydrocephalus

usually exhibits a thin rim of compressed cerebral cortex and an absent falx cerebri only in extreme cases. The fusion of the cerebral hemispheres and basal ganglia, the lack of an interhemispheric fissure, and frequently facial abnormalities- all of which were absent in our patient- are characteristics of alobar holoprosencephaly. Porencephaly is the term for cystic cavities that form in the cerebral hemispheres due to an infection or infarction; unlike hydranencephaly, some normal brain parenchyma is usually maintained. Therefore, the key to validating hydranencephaly in this case was imaging findings, particularly the full replacement of cerebral hemispheres with CSF, retained midline structures, and normal posterior fossa anatomy.

Recent literature has documented occasional occurrences of hydranencephaly, emphasising the diagnostic variability and imaging range. Zhang H et al., (2025) examined CT findings in 22 instances and emphasised the diagnostic relevance of detecting preserved midline and posterior fossa structures, which is consistent with our observations [9]. Toumi K et al., (2024) also described a newborn case and emphasised the relevance of prenatal imaging in early diagnosis [10], whereas Sharma N et al., (2021) published a case with partial cortical tissue preservation, illustrating that hydranencephaly can occur at varying degrees of severity [11]. Compared to earlier studies, our case is unique because of the postnatal onset of seizures, the lack of prenatal detection, and the specific CT findings indicating total bilateral hemisphere loss with spared brainstem and cerebellum.

The primary objectives of palliative and supportive care are to manage symptoms associated with hydranencephaly. Dietary support, anticonvulsant medications, and the management of infections or hydrocephalus are alternative options. Although these operations do not alter the overall prognosis, ventriculoperitoneal shunting or endoscopic choroid plexus cauterisation may be considered in certain instances to reduce head size and intracranial pressure. Multidisciplinary follow-up is essential [2].

This case is being reported because of its rarity and distinct clinical and imaging presentation. The combination of postnatal seizure onset, failure to thrive, and the extensive imaging findings makes this report a valuable addition to existing literature on hydranencephaly.

CONCLUSION(S)

Hydranencephaly is an uncommon and debilitating congenital brain deformity that can manifest neurological symptoms such as convulsions, sleepiness, and failure to thrive after the newborn period. The diagnosis in our instance was primarily made possible through neuroimaging, as CT results showed that the deep brain structures were preserved and the cerebral hemispheres were almost absent. This instance highlights the need to consider structural brain abnormalities such as hydranencephaly when treating babies with unexplained seizures and developmental delays.

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