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Case Report

Hydranencephaly: a rare case report on enlarging head in an infant

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ABSTRACT

Hydranencephaly is an uncommon congenital disorder in which the cerebral hemispheres are absent and replaced by cerebrospinal fluid. In this article, we describe an uncommon case of hydranencephaly in a 3-month-old male child with complaints of progressively increasing head size over the last 1 month and inability to control his neck. The child was diagnosed as a case of Hydranencephaly by cranial ultrasonography (USG) and further confirmed by computed tomography (CT) and magnetic resonance imaging (MRI) which revealed a complete absence of the cerebral hemisphere with intact falx, brain parenchyma was replaced by a sac-like structure containing CSF, reduced volume of the midbrain, and a thin rim of brain tissue was seen at the frontal and temporal region bilaterally and at the lateral aspect of the falx cerebri. The child was managed using a ventriculoperitoneal shunt procedure. Hydranencephaly requires prompt diagnosis using Cranial USG, CT, or MRI and treatment by ventriculoperitoneal shunt procedure and a multi-disciplinary approach. Proper parental counselling is required as it has a bad prognosis.

Keywords: Computed tomography, Congenital cerebral malformations, Cranial ultrasound, Hydranencephaly, Hydrocephalus, Magnetic resonance imaging, Ventriculoperitoneal shunt

INTRODUCTION

Hydranencephaly is a rare congenital disorder of the central nervous system in which cerebrospinal fluid (CSF) completely replaces the cerebral hemispheres, but the falx cerebri, thalamus, and cerebellum are intact.[1,2] It can be distinguished from other CNS anomalies by normally appearing facial features and midline structures.[3] It affects less than 1 per 10,000 births, 0.2% of autopsies of infants, and approximately 1% of clinically diagnosed cases of hydrocephalus in infants.[4] Clinical differentiation of hydranencephaly and extreme hydrocephalus is a must, as the latter carries a better prognosis.[4] Here, we report a rare hydranencephaly case seen in a rural tertiary care setting.

CASE REPORT

Chief complaints

A 3-month-old male infant presented to our pediatrics outpatient department (OPD) at Hind Institute of Medical Sciences, Sitapur with complaints of progressive increase in head size over the last 1 month and inability to control his neck.

Detailed history

A detailed history was taken from the parents. This infant was born of third-degree consanguineous marriage, second in birth order with a maternal age of 24 years and paternal age of 25 years at the time of conception. There was a history of spontaneous abortion 1.5 years back due to an unknown cause at 2 months of gestation. This pregnancy was confirmed at home by urine pregnancy test kit at 3 months of gestation. The mother had not registered for the antenatal care clinic. There was no history of fever with rash, radiation exposure, or exposure to butane gas or carbon monoxide gas during this pregnancy in the mother. This G3 P1 L1 A1 (gravida 3, parity 1, live 1, abortion 1) unbooked mother presented in our obstetric emergency in labour at 37 weeks of gestation. Cardiotocography of the fetus was normal with no remarkable clinical obstetric complications as per her

documents and the mother delivered at our tertiary care center through vaginal delivery. The birth weight of the baby was 2400 grams, length was 45 cm, and head circumference (HC) was 35 cm as per documents. The baby did not cry immediately after birth and neonatal resuscitation for more than one minute was carried out. The baby was admitted to the neonatal intensive care unit (NICU) and intubated for three days requiring ventilator support. He had an episode of abnormal body movement during his NICU stay. The baby was discharged after 10 days of NICU stay and advised phenobarbitone on discharge as per the documents and was later lost to follow-up. The baby's weight at discharge was 2460 grams, length was 45 cm and HC was 35 cm.

Examination

On examination at 3 months on his presentation, the child had delayed developmental milestones, a grossly enlarged head size with a head circumference of 46 centimeters (cm) that was > 97th percentile according to the WHO growth chart for 0-5 years boys, and craniofacial disproportion (Figure 1). The anterior fontanelle measured 4×4 cm, bulging, tense, and cracked pot sign was present. Posterior fontanelle enlargement with diastasis of sutures was present. On ophthalmic examination, the setting sun sign was present and pupils were equally normal sized and normal reaction. Primitive reflexes like sucking reflexes were intact. On detailed CNS examination, the tone in all four limbs and deep tendon reflexes were normal. Examination of the other systems along with the spine was normal.

Investigations

On investigations, hemoglobin was 9.3 gram/deciliter, total leukocyte count was 9200 per cubic millimeter (cm), and platelets were 4.22 lakhs/cm. The C-reactive protein was negative. The liver function tests, kidney function tests, serum electrolytes, and CSF cyto-biochemical analysis were within the normal range. Blood and CSF culture sensitivity were sterile. The TORCH profile and genetic testing could not be done due to financial constraints in this case.

Cranial ultrasound

The infant was provisionally diagnosed as hydrocephalus clinically and was evaluated for the same by ultrasonography (USG) of the cranium. USG cranium revealed severe dilatation of the frontal horn of the lateral ventricle and third ventricle with thin corpus callosum, and effacement of sulci and gyri (Figure 2).

Non-contrast computed tomography scan

A plain NCCT scan of the head was done to evaluate further. The NCCT Head showed a complete absence of the cerebral hemisphere with intact falx (Figure 3).

Magnetic resonance imaging

MRI brain revealed a large hypointense sac-like structure isointense to CSF with near complete loss of cortical brain matter and cerebral hemisphere (Figure 4). Also, the midbrain appeared reduced in volume with intact Falx cerebri.

Management

The neurosurgeon advised ventriculoperitoneal shunting. Parents were explained about the grave prognosis and possible complications. The child underwent a ventriculoperitoneal shunt procedure and the general condition of the baby improved along with the reduction in head size. Parents received psychological support and had frequent follow-up appointments to monitor and treat any issues that may arise.

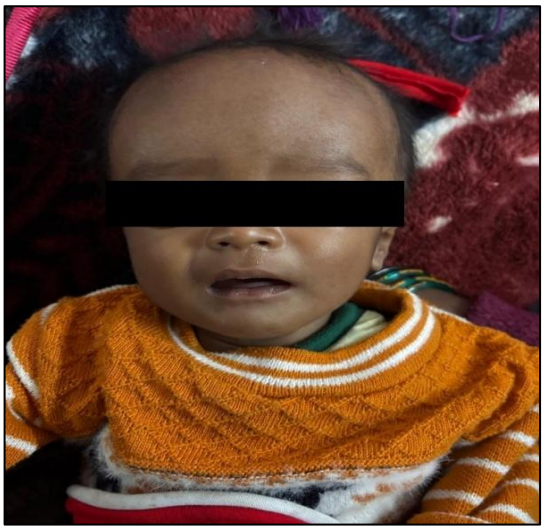

Figure 1: Grossly enlarged head size of the infant at the time of presentation (3 months age).

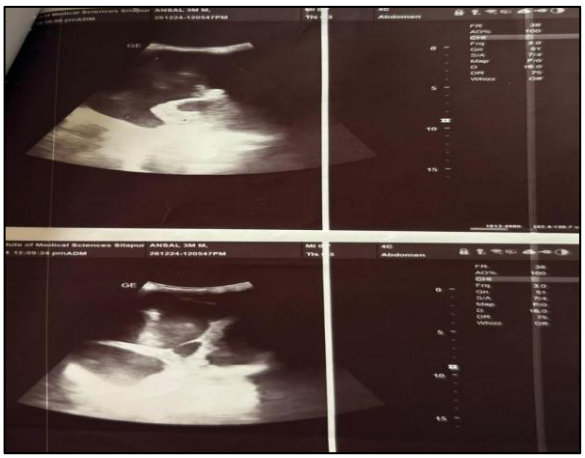

Figure 2: USG cranium showing severe dilatation of frontal horn of lateral ventricle and third ventricle with thin corpus callosum, effacement of sulci and gyri.

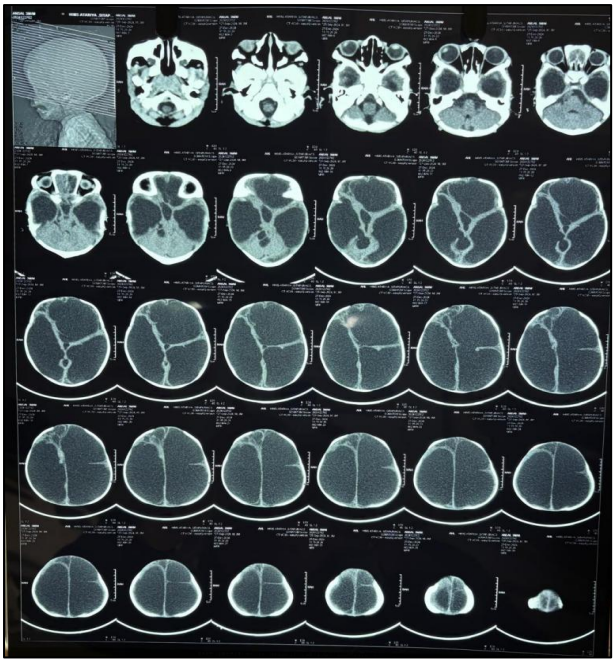

Figure 3: NCCT head shows complete absence of cerebral hemisphere with intact falx, brain parenchyma is replaced by a sac-like structure containing CSF, and a thin rim of brain tissue was seen at the frontal and temporal region bilaterally and at the lateral aspect of the falx cerebri.

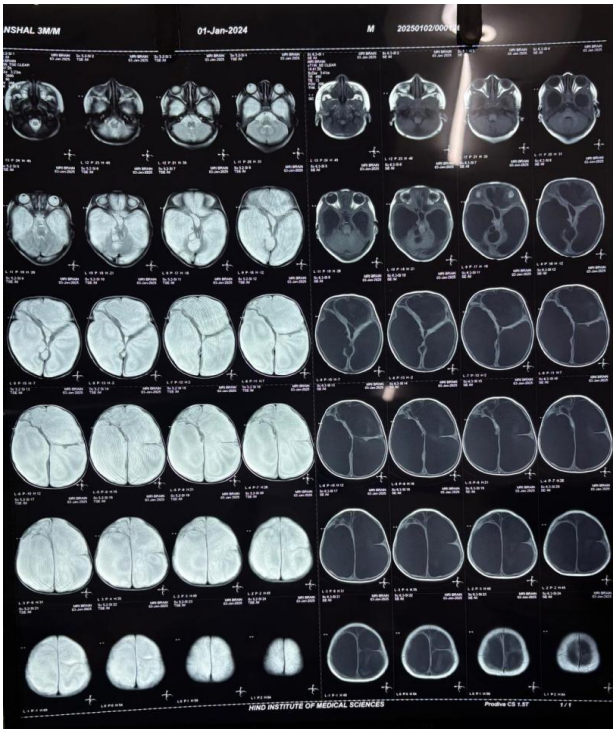

Figure 4: MRI BRAIN reveals a large hypo-intense sac-like structure isointense to CSF with near complete Loss of cortical brain matter and cerebral hemisphere with intact falx cerebri and reduced volume of the midbrain suggestive of hydranencephaly.

DISCUSSION

Hydranencephaly is a rare encephaloclastic disorder characterized by the loss of cerebral hemispheres, which are replaced by cerebral spinal fluid and necrotic debris, surrounded by leptomeninges.[5] Usually the cerebral cortex is not present, although a section of the occipital lobe can at times be intact.[6] Within the skull, structures including the cerebellum, brainstem, choroid plexus, thalamus, midbrain, and basal ganglia are usually intact.[6] The falx cerebri is normally present, however it can be partly or entirely missing, as well as the septum pellucidum.[4] Likewise, in the present case, the brain structures other than the cerebral cortex were still visible with the midbrain appearing reduced in volume, and the Falx cerebri appeared intact.

Various etiopathogenesis theories have been postulated to describe the heterogeneity of its occurrence. The most prevalent etiology identified is an obstruction of the supra-clinoid portion of the bilateral internal carotid arteries, which causes ischemic degeneration of the structures that are supplied by them.[4]

Localized damage to brain tissues leading to hydranencephaly can be caused due to viral infections like adenovirus, cytomegalovirus, enterovirus, Epstein-Barr virus, herpes simplex virus, parvovirus, respiratory syncytial viruses, and intrauterine infections, such as congenital toxoplasmosis.[7] Maternal exposure to butane gas or carbon monoxide has also been identified as an etiology that can cause fetal hypoxia followed by extensive tissue necrosis with cavitations, necrotized tissue resorption, and necrotizing vasculitis.[8] Trisomy, arthrogryposis, renal aplastic dysplasia, poly-valvular heart disease, and Fowler syndrome are among the congenital conditions that have been linked to hydranencephaly.[9]

The possible etiology in our case can be due to severe perinatal asphyxia as these cases of hypoxic-ischemic brain injury from birth asphyxia can lead to widespread destruction of brain tissue, resulting in the typical features of hydranencephaly where most of the cerebral hemispheres are absent and replaced by cerebrospinal fluid, essentially "melting away" the brain tissue due to severe damage. Cranial USG of the present case revealed severe dilation of the frontal horn of the lateral ventricle and third ventricle with thin corpus callosum, and effacement of sulci and gyri thereby giving the impression of hydrocephalus. Hydranencephaly is diagnosed by cranial ultrasonography, which reveals a massive cystic mass occupying the whole intracranial cavity with the cerebral cortex absent or discontinuous.[4,10]

Along with a midline echo from the remains of the falx, the tentorium cerebelli, the cerebellum, the thalami, and the brainstem extending into the cystic space are distinctive features. A single ventricle in the middle may

result from the lack of the septum pellucidum, and the third ventricle and choroid plexus were frequently visible.[4,10] The diagnosis of porencephaly, alobar holoprosencephaly, and severe hydrocephaly differs significantly.[11,12]

The aforementioned structures will still be encircled by a cortical rim in these circumstances; it is important to attempt to differentiate them during hydranencephaly ultrasonographic imaging since they have a better prognosis.[11,12] In severe cases of hydrocephalus, magnetic resonance imaging (MRI) or intrauterine CT scans are crucial for validating the diagnosis since the thin cortical layer can be challenging to detect on sonography.[4]

In the present case, the NCCT brain study revealed that there is evidence of complete absence of the cerebral hemisphere with falx intact, brain parenchyma is replaced by a sac-like structure containing CSF, and a thin rim of brain tissue was seen at the frontal and temporal region bilaterally and at the lateral aspect of the falx cerebri. Posterior fossa contents including the cerebellar hemispheres, vermis, and fourth ventricle were normal in our case with intact bony calvarium and normal pericalvarial soft tissues.

These findings of NCCT brain gave an impression of hydranencephaly in our case. Our study findings were consistent with different studies.[4,13] MRI brain of our case revealed evidence of a large hypointense sac-like structure isointense to CSF with near complete loss of cortical brain matter and cerebral hemispheres with intact Falx cerebri and reduced volume of the midbrain. There was no evidence of abnormal area of restricted diffusion on Diffusion-weighted imaging (DWI) or susceptibility blooming on gradient images noted. The cerebellum and the rest of the brain stem appear normal. These findings were consistent with the diagnosis of Hydranencephaly. Our MRI brain findings were consistent with different studies.[14]

In the present case, the child underwent a ventriculoperitoneal shunt procedure and the general condition of the baby improved. In children with hydranencephaly managed by ventriculoperitoneal shunt (VPS) insertion, endoscopic choroid plexus coagulation (CPC), open choroid plexectomy (CPlx), the head circumference reduced by 1 cm or more in comparable proportions (62–71%), and increased in those who got palliative care.[15] At one year of age, babies treated with CPlx had a lower mortality rate (43%) compared to those managed through VPS, CPC, or palliative care (70-82%).[15]

Most documented instances of hydranencephaly had remnant or retained midbrain structures, with the thalamus or basal ganglia (or both) areas retained. Brainstem functions are majorly affected thus newborns with hydranencephaly have a grave prognosis. Although the majority of them pass away before birth, the first year of life is when the greatest mortality occurs.[4,16] Even if a child survives, they will undoubtedly have significant disabilities.[4,16]

CONCLUSION

The most likely consequence based on the ultrasonographic data we reported was hydranencephaly, which was verified by head CT and MRI. Early diagnosis and timely intervention are crucial in hydranencephaly. Appropriate counseling of the parents is a must due to its poor prognosis. Multi-disciplinary approach to the management of hydranencephaly is needed.

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